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(54) Title: CHARACTERIZATION OF SINGLE NUCLEOTIDE POLYMORPHISMS IN CODING REGIONS OF HUMAN GENES		
(57) Abstract The invention provides nucleic acid segments of the human genome, particularly nucleic acid segments from the coding region of a gene, including polymorphic sites. Allele-specific primers and probes hybridizing to regions flanking or containing these sites are also provided. The nucleic acids, primers and probes are used in applications such as phenotype correlations, forensics, paternity testing, medicine and genetic analysis.		

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CHARACTERIZATION OF SINGLE NUCLEOTIDE POLYMORPHISMS
IN CODING REGIONS OF HUMAN GENES

RELATED APPLICATION

This application claims the benefit of U.S. Provisional Application Serial
5 No. 60/127,248, filed March 31, 1999, the entire teachings of which are incorporated
herein by reference.

GOVERNMENT SUPPORT

The invention was supported, in whole or in part, by grant 5-P50-HG00098-
09 SNP from the National Institutes of Health (NCHGR) and grant 1-R01-
10 HL61774-01 from the National Institutes of Health (NHLBI). The Government has
certain rights in the invention.

BACKGROUND OF THE INVENTION

A major goal in human genetics is to understand the role of common genetic
variants in susceptibility to common diseases (N. Risch and K. Merikangas, *Science*,
15 273:1516 (1996.); *E. S. Lander, Science*, 274:536 (1996); F.S. Collins, *et al.*,
Science, 278:1580 (1997)). This requires assembling an extensive catalogue of
single-nucleotide polymorphisms (SNPs) and performing systematic association
studies for particular diseases.

The human population has relatively limited genetic diversity, reflecting its
20 young age and historically small size (F. J. Ayala *et. al.*, *Proc. Natl. Acad. Sci.*,
91:6787 (1994)). Given the restricted nature of the allelic spectrum, some authors
have recently suggested that it should eventually be possible to collect all common
SNPs in the human population and have hypothesized that such common variants
may underlie much of the genetic risk of common disease (N. Risch and K.
25 Merikangas, *Science*, 273:1516 (1996.); *E. S. Lander, Science*, 274:536 (1996); F.S.
Collins, *et al.*, *Science*, 278:1580 (1997)). This is in contrast to the situation for rare

Collins, *et al.*, *Science*, 278:1580 (1997)). This is in contrast to the situation for rare genetic diseases, which are primarily caused by a large number of distinct alleles that are recent, rare and highly penetrant. Important examples of associations to common (>1%) alleles include the ApoE4 allele in Alzheimer's disease, the Factor
5 V^{Leiden} allele in deep-venous thrombosis, and the CCR5-Δ32 in resistance to HIV infection (A. M. Saunders *et al.*, *Neurology*, 43:1467 (1993); R. M. Bertina, *Nature*, 369:64 (1994); M. Dean *et al.*, *Science*, 273:1856 (1996)). The most relevant variants are likely to be those in coding and regulatory regions of genes.

SUMMARY OF THE INVENTION

10 As described herein, the nature of SNPs in the coding regions of human genes has been explored. SNPs were identified in 106 genes relevant to cardiovascular disease, endocrinology and neuropsychiatry, by screening an average of 114 independent alleles using two independent screening methods. To ensure high accuracy, all reported SNPs were confirmed by DNA sequencing. A total of
15 545 SNPs were identified, including 395 coding-regions SNPs (cSNPs) divided roughly equally between those causing synonymous and non-synonymous changes. The cSNPs most likely to influence disease, those that alter the amino acid sequence of the encoded protein, show strikingly different properties: they occur at a lower rate and with lower allele frequencies. This likely reflects selection acting against
20 deleterious alleles during human evolution. The lower allele frequency of cSNPs has important implications for the number of chromosomes that must be sampled to construct a comprehensive catalogue of human cSNPs.

The invention relates to a gene which comprises a single nucleotide polymorphism at a specific location. In a particular embodiment the invention
25 relates to the variant allele of a gene having a single nucleotide polymorphism, which variant allele differs from a reference allele by one nucleotide at the site(s) identified in Figures 5A-5Q. Complements of these nucleic acid segments are also included. The segments can be DNA or RNA, and can be double- or single-stranded. Segments can be, for example, 5-10, 5-15, 10-20, 5-25, 10-30,
30 10-50 or 10-100 bases long. The invention further relates to gene products encoded by genes and oligonucleotides of the invention.

The invention further provides allele-specific oligonucleotides that hybridize to a gene comprising a single nucleotide polymorphism or to the complement of the gene. These oligonucleotides can be probes or primers.

The invention further provides a method of analyzing a nucleic acid from an individual. The method determines which base is present at any one of the polymorphic sites shown in Figures 5A-5QQQQQQQ. Optionally, a set of bases occupying a set of the polymorphic sites shown in Figures 5A-5QQQQQQQ is determined. This type of analysis can be performed on a number of individuals, who are tested for the presence of a disease phenotype. The presence or absence of disease phenotype is then correlated with a base or set of bases present at the polymorphic site or sites in the individuals tested.

BRIEF DESCRIPTION OF THE DRAWINGS

Figure 1 is a graph showing minor allele frequency by polymorphism type. The percentage of cSNPs having minor allele frequency classified as low (<5%), medium (5-15%) or high (>15%) frequency is displayed for synonymous, non-synonymous and non-coding SNPs.

Figure 2 is a graph showing the distribution of nucleotide diversity. Normalized frequency of variant sites, θ , was calculated for the coding region of each gene. The graph shows the percentage of genes having θ in the indicated range.

Figures 3A and 3B are a table showing a summary of polymorphisms in 106 human genes described herein. Column 1 shows the name of the gene as used in Online Mendelian Inheritance in Man. Column 2 shows the number of coding base pairs screened. Column 3 shows the number of synonymous (or silent) polymorphisms identified. Column 4 shows the number of non-synonymous polymorphisms identified. Column 5 shows the number of non-coding base pairs screened. Column 6 shows the number of non-coding polymorphisms, including those in introns and untranslated regions (UTR), identified.

Figure 4 is a table showing polymorphism rates for different classes of sites.

Figures 5A-5QQQQQQQ are a table showing the specific polymorphisms identified in the genes studied as described herein. Column 1 shows the laboratory

designation for the polymorphism. Column 2 shows the name of the gene as used in Online Mendelian Inheritance in Man. Column 3 shows the reference nucleotide which occupies the polymorphic site in the reference allele. Column 4 shows the variant nucleotide which occupies the polymorphic site in the variant allele.

- 5 Column 5 shows the reference amino acid encoded by the codon which contains the polymorphic site in the reference allele. Column 6 shows the variant or alternate amino acid encoded by the codon which contains the polymorphic site in the variant allele. Column 7 indicates whether the polymorphism is located in the coding or non-coding region of the gene. Column 8 shows the assay number in which the
- 10 polymorphism was assessed. Columns 9 and 10 show the forward and reverse primers, respectively, which were used to identify the polymorphism. Column 11 shows the sequence of the gene used in the assay, with the polymorphic site indicated by brackets and the primers shown in capital letters. Column 12 shows the total number of nucleotides given in Column 11.

15 DETAILED DESCRIPTION OF THE INVENTION

- There is a rich literature concerning nucleotide variation in model systems, particularly in *Drosophila* (E. N. Moriyama and J. R. Powell., *Mol. Biol. Evol.*, 13:261 (1996)), but sequence variation in human genes has been studied only in limited ways. A small number of studies have focused on individual genes (such as
- 20 beta-globin and lipoprotein lipase) in many individuals, and one study examined 49 genes by comparing two independent sequences deposited in public databases (R. M. Harding *et. al.*, *Am. J. Hum. Genet.*, 60:772 (1997); D. A. Nickerson *et. al.*, *Nature Genetics*, 19:233 (1998); W. H. Li and L.A. Sadler, *Genetics* 129:513 (1991)). To perform a more comprehensive survey, as described herein, a collection
- 25 of 106 genes were selected whose protein products play important roles in the cardiovascular, endocrine and neurological systems (Figures 3A-3B and Figures 5A-5Q). Gene sequences were obtained from the Genbank and TIGR databases. Where multiple sequence depositions were available, a consensus sequence was derived. Determination of coding sequence, untranslated regions and
- 30 intronic regions was based on annotation in the public database, although internal

checks were performed to ensure accurate determination of start and stop codons, open reading frames and the like.

The genes were chosen because of their relevance to common, clinically significant diseases, such as coronary artery disease, diabetes, and schizophrenia.

5 They encode proteins involved in coagulation, lipid metabolism, energy metabolism, neuroendocrine physiology, neurotransmission and central nervous system development. Variation in these genes was studied in a sample including Caucasians, African-Americans, African Pygmies and Asians, with an average of 114 chromosomes screened for each gene. Of the samples screened, 30 were from
10 Caucasian individuals, 14 from Asian, 10 African American and 7 Africans. The average number of individuals successfully screened for each gene was 57, with the precise number successfully screened varying among genes. Cell lines were obtained from Coriell Cell Repository, and DNA prepared according to standard protocols. In addition, 10 of the Caucasian samples used in this study were obtained
15 as anonymous blood samples from the Physician's Health Study (gift of Charles Hennekens and J. Michael Gaziano). The sample size provides greater than 65% power to detect alleles with frequency of 1%.

Overall, the sample of 114 chromosomes was screened for SNPs in a total of 195.4 kb, consisting of 135.8 kb of coding regions and 59.6 kb from adjacent
20 non-coding region (untranslated region (UTR) and introns). Sequences were amplified by the polymerase chain reaction (PCR) and screened by two independent methods. The first method involved hybridization of labeled PCR products to variant detector arrays (VDAs) (that is, high density DNA probe arrays containing oligonucleotides specific for the sequences under study) (M. Chee *et al.*, *Science*,
25 274:610 (1996); D. G. Wang *et al.*, *Science*, 280:1077 (1998)); variant sequences typically give rise to altered hybridization patterns. These chips contained variant detector arrays (VDA) (M. Chee *et al.*, *Science* 274:610 (1996)).

Using VDAs, candidate SNPs were identified using a combination of three algorithms followed by visual inspection. For each base position and strand queried
30 there are four VDA features: one contains the expected base (the reference sequence) in the central position and the other three features contain central substitution bases (in the background of the reference sequence). The base-calling

algorithm looked for positions at which hybridization to a substitution base gives a stronger signal than the reference base. The second algorithm (mutant fraction) examined the reference base and each one of the substitution bases in turn and calculates the fraction of signal present in the non-reference base. The final
5 algorithm (footprint detection) depends upon a loss of signal at the reference positions surrounding a nucleotide substitution. These algorithms are combined to yield a confidence score of "certain" or "likely" for each candidate polymorphism. Two analysts independently scored the data, and candidate polymorphisms found by either observer were included in subsequent confirmation tests. PCR assays
10 spanning each exon were designed using Primer 3.0 release 0.7. PCR was performed according to standard protocols, and assays destined to be hybridized to the same chip design were pooled together. Chip samples were prepared and hybridized as described in D.G. Wang *et al.* (*Science* 280:1077 (1998)), except that pools consisting of about 100 assays contained 5-6 µg of amplified material. In all,
15 854 assays (average size of 300 bp, covering 106 genes) were amplified from each individual and were hybridized to 12 distinct chip designs. The probe arrays were designed to query only the coding sequence for some genes, while other genes contained the entire mRNA and/or surrounding intron (Figures 3A-3B). The second method involved subjecting PCR products to Denaturing HPLC (dHPLC) (P. J.
20 Oefner and P. A. Underhill, *Am. J. Hum. Genet.*, 57:A266 (1995)) at a critical temperature; heterozygous individuals typically give rise to heteroduplex products with altered denaturation and migration properties.

Sequences were amplified as above except that the final extension in the PCR protocol was followed by denaturation and slow reannealing to allow
25 heteroduplex formation. A total of 6 µl of each individual PCR product was injected into Wave DNA Fragment Analysis System (Transgenomic). A total of 592 of the VDA assays (covering the 89 genes attempted with this method) were successfully screened by DHPLC. Only assays of >160 base pairs were used for DHPLC, because shorter assays performed unreliably for mutation detection. The
30 DHPLC parameters (percentage of acetonitrile, column temperature) used for each fragment were automatically calculated using a novel predictive algorithm, and DHPLC traces were analyzed using the clustering program ASH v2.0. A scoring

algorithm was developed based upon the similarity score by ASHv2.0 and contour of the elution profile.

Because both screening methods can generate to a significant number of false positives, it was important to confirm every reported SNP. Samples implicated
5 by either method as containing a candidate SNP were thus subjected to fluorescent dideoxy sequencing, either to confirm the presence of the SNP (in the case of the chip) or to identify and confirm the presence of the SNP (in the case of DHPLC). Such confirmation proved essential for eliminating false positives.

Candidate SNPs were either validated (if found by VDAs) or identified (if
10 implicated by DHPLC) by DNA sequencing. For this purpose, sequences were amplified with PCR primers tailed with standard M13 sequencing sites (-21 forward and -28 reverse) and conventional dye-primer sequencing was performed on ABI 377 sequencers. For candidate SNPs discovered by VDAs, one individual was chosen (a candidate homozygous variant, when available, or a candidate
15 heterozygote) and sequencing was performed on one strand to confirm by visual inspection the presence of the SNP at the indicated position. For amplicons found to be polymorphic by DHPLC, two individuals were selected representing each distinct elution pattern observed and were sequenced on both strands to discover the variant base or bases. Sequences were base-called by the Phred program, assembled
20 by the Phrap program, and polymorphism candidates were identified by the PolyPhred program (D. A. Nickerson *et. al.*, *NAR*, 25:2745 (1997)). All results were visually inspected by at least two observers.

The overall false positive rate for VDAs was 45%. The rate was much lower (about 10%) for certain chip designs, synthesis protocols, and for candidate
25 polymorphisms scored as "certain." The false positive rate among fragments displaying an altered elution pattern by DHPLC was similar (40%). The false positive rates reflect the thresholds employed for declaring a candidate SNP, which were chosen to ensure high sensitivity.

A total of 545 SNPs were identified in the 195 kb surveyed, consisting of
30 150 non-coding SNPs and 395 cSNPs. Results from these studies are shown in the Figures. The complete data are available on the web site
http://www.genome.wi.mit.edu/cvar_snps; access to this website can be gained

using the guestname "snp_pilot" and the password "noynek". In the future, access to this website may be available to the public, and thus, no guestname or password may be needed.

To directly determine the false-negative rate of the screen, conventional
 5 DNA sequencing was performed on ten of the genes (THPO, TBAX2R, PTHLH, IGF2, HTR2A, HTR1A, GHR, GABRB1, F10, and CYP11B1) spanning 25.2 kb in twenty individuals. Sequencing was performed on both strands using dye-primer chemistry and sequence traces were interpreted using PolyPhred (D.A. Nickerson *et al.*, *NAR*, 25:2745 (1997)). VDA analysis identified 85% of variants found by direct
 10 sequencing, while DHPLC identified 87% of the variants found by direct sequencing. In regions screened by both VDAs and DHPLC, the combination of the two methods identified 100% of the polymorphisms found by direct sequencing.

Overall, about one-third of individuals were screened with both methods, and one-third were screened with each of the two methods alone. (For some genes, the
 15 non-coding regions were screened only by DHPLC.) It is estimated that the false negative rate over the entire study to be about 15% for regions screened by one method, and negligible for sequences screened by both methods. The total number of true polymorphisms not identified is estimated to be less than 10%.

A SNP survey can be characterized in terms of either K, the observed
 20 number of variant sites, or p, the observed heterozygosity per bp. Because K increases with the number of chromosomes (n) studied and the total sequence length L, it is preferable to use the normalized number of variant sites

$$\hat{\theta} = K / \left(\sum_{i=1}^{n-1} i^{-1} \right) L \text{ which corrects for sample size. Under the neutral}$$

theory of molecular evolution and infinite sites model, θ and π are both estimators
 25 of the population genetic parameter $\theta = 4N\mu$ (Li, *Molecular Evolution*, Sinauer Associates (1997), Canada).

SNPs were found at a similar overall frequency in coding and non-coding regions. SNPs in coding region occurred at a frequency of 1 per 344 bp, corresponding to $\hat{\theta} = 5.47 \times 10^{-4}$ and $\pi = 5.07 \times 10^{-4}$. Interestingly, SNPs were
 30 observed in non-coding DNA at a similar frequency of 1 per 397 bp. The

normalized number of variant sites was $\theta = 4.93 \times 10^{-4}$, and the mean heterozygosity (π) = 5.05×10^{-4} (Figure 4). Calculations of π involve allele frequencies. Polymorphisms identified by DHPLC alone were excluded because we did not sequence all of the samples showing a variant DHPLC pattern and thus could not be certain of allele frequency. The estimates of π were thus based on 411 of 545 polymorphisms. Although the VDAs were designed for polymorphism discovery rather than genotyping, the estimated allele frequencies proved to be quite accurate. Specifically, genotyping assays (employing single-base extension assays) for 25 SNPs yielded allele frequencies that differed by an average of only 2% from those estimated on the basis of genotypes inferred from the VDA. For both classes, the similar values for θ and π is consistent with a population evolving according to neutral expectations.

The 395 cSNPs were roughly equally divided between synonymous (203 cSNPs) and non-synonymous (192 cSNPs) changes. Since approximately two-thirds of random mutations would alter an amino acid, the fact that non-synonymous cSNPs comprise slightly less than half of the cSNPs implies strong selection against amino-acid altering changes. To address this issue more directly, the nucleotide diversity was examined at four-fold degenerate sites, two-fold degenerate sites, and non-degenerate sites. Changes at four-fold degenerate sites produce only synonymous changes, while those at non-degenerate sites are always non-synonymous. Nucleotide diversity (θ) was 9.64×10^{-4} at four-fold degenerate sites, 6.85×10^{-4} at two-fold degenerate sites, and 3.70×10^{-4} at non-degenerate sites. Assuming that mutations occur at an equal rate at both classes of sites, non-synonymous variants survive to be detected in such a survey at only 38% of the rate of synonymous changes.

The force of selection is also evident in comparing non-synonymous cSNPs causing a non-conservative amino acid alteration with those causing a conservative amino-acid change. Conservative and non-conservative amino acid substitutions were defined for this analysis according to the BLOSUM62 matrix, used in sequence comparison (S. Henikoff and J. G. Henikoff, *PNAS*, 89:10915 (1992)). Conservative changes were those having a positive or neutral sign in the matrix, while non-conservative changes were those having a negative value. Non-conservative

- cSNPs represent only 36% of the non-synonymous cSNPs, whereas randomly distributed mutations would be expected to produce a higher proportion (52%) of non-conservative changes. The proportion of non-synonymous SNPs expected to cause a non-conservative amino acid substitution was determined based on the actual codon usage in the 106 genes studied, the known frequencies of transitions and transversions, and the definition of non-conservative changes employed in the BLOSUM62 matrix. This implies that non-conservative cSNPs survive to be detected in such a survey at only about half of the rate of conservative, non-synonymous cSNPs.
- 10 The various types of SNPs differ not only in the rate of their occurrence, but also in the frequency of their minor alleles. This can be seen in several ways. When SNPs are classified according to whether the frequency of the minor allele was high ($\geq 15\%$), intermediate (5-15%) or low ($\leq 5\%$), it is clear that the non-synonymous cSNPs were enriched in low frequency alleles compared to the rest of the collection
- 15 (Figure 1). The distribution of non-synonymous allele frequencies was significantly different than that of synonymous changes ($p=0.02$, Kolmogorov-Smirnov test). Indeed, more than half (58%) of non-synonymous cSNPs were found at a frequency below 5%, with this effect evident for both conservative and non-conservative substitutions.
- 20 The effect of selection can also be inferred by considering the average frequency of the minor allele: it is 8% for non-conservative cSNPs, 11% for conservative but non-synonymous cSNPs, and 14% for both synonymous cSNPs and non-coding SNPs. In addition, the lower allele frequency of non-synonymous cSNPs is reflected in the fact that the heterozygosity π is lower than the normalized
- 25 rate of variant sites $\hat{\theta}$ for this class of SNPs (Figure 4). This divergence is in the direction predicted by the action of purifying selection, although it falls short of statistical significance. Tajima's D was non-significant. (F. Tajima, *Genetics*, 123:545 (1989).
- 30 The distribution of SNPs among the 106 genes was explored, with an eye toward detecting differential effects of selection among genes. The number of cSNPs per gene ranged from 37 for Factor V to 0 for thirteen of the genes, and the normalized rate, $\hat{\theta}$, similarly showed considerable variation (Figure 2). The

- observed variation in nucleotide diversity is similar in magnitude to that observed for *Drosophila* (E. N. Moriyama and J. R. Powell., *Mol. Biol. Evol.*, 13:261 (1996)). Variation among genes could be due to many factors (D. J. Begun and C. F. Aquadro, *Nature*, 356:519 (1993); Nachman *et. al.*, *Genetics*, 150:1133 (1998)).
- 5 The fact that non-synonymous cSNPs show a somewhat wider variation than synonymous cSNPs (the coefficient of variation is 20% larger for the former class) is consistent with differences in selective constraints among loci, but the difference falls well below statistical significance. A variety of population genetic tests are available for testing selection at individual loci (M. L. Wayne and K. L. Simonson,
- 10 *Trends and Ecology and Evolution*, 13:236 (1998)).

The age of a SNP allele has important implications for its use in human genetic studies. Recently-occurring SNP alleles are more likely to show extensive linkage disequilibrium (retention of the ancestral haplotype on which they arose) as compared to older SNPs. Such linkage disequilibrium can provide a powerful tool

15 in identifying disease genes (E. S. Lander, N.J. Schork, *Science*, 265:2037 (1994)). Although the precise age of the SNPs could not be assessed from these studies, characterization of which allele preceded human speciation and which arose thereafter was sought. To determine the ancestral human allele, each corresponding gene was sequenced from the common chimpanzee (*P. troglodytes*). Each assay

20 used in the human survey was amplified from a single chimpanzee (DNA gift of Kristin Ardlie) and subjected to dye-primer sequencing on both strands. A single chimpanzee sample will accurately reveal the ancestral allele except in cases where the site has mutated and fixed during the chimpanzee evolution or is polymorphic in the chimpanzee population and happened to be homozygous for the non-ancestral

25 allele. These two cases are quite rare (probably less than 2%) and thus have been neglected for the purpose of estimating overall rates. A human allele was considered to be ancestral if it was present in the homozygous state in the chimpanzee sample. A total of 136 kb of chimpanzee sequence was obtained, revealing an inter-species divergence of 0.6% in the regions studied.

- 30 An elegant result in theoretical population genetics predicts that the probability that a neutral allele represents the ancestral state should be equal to its frequency in the population (G. A. Watterson and H. A. Guess, *Theoretical*

Population Biology, 11:141 (1977)). The minor allele should thus represent the ancestral state in a predictable proportion of cases. The ancestral allele and minor allele frequency was determined for 267 of the reported SNPs. For 3 of the 267 SNPs, the chimpanzee was homozygous for a third allele differing from both of the
5 current human alleles. This is consistent with the overall 0.6% nucleotide sequence divergences seen between human and chimpanzee. Among polymorphisms with a minor allele frequency below 10%, the average allele frequency was 3% and the proportion that was ancestral was 7% (11/158) of cases. Among polymorphisms with minor alleles exceeding 10%, the mean frequency was 28% and the proportion
10 that were ancestral was 32% (35/109). These results thus agree remarkably well with the theoretical prediction, providing the first reported test of this prediction in humans. It therefore follows that the minor SNP allele need not be the younger allele; this has implications for linkage disequilibrium mapping.

The distribution of SNPs among Caucasian, African-American, African and
15 Asian samples was also examined. Although the vast majority of SNPs were seen in multiple groups, there was a statistically significant excess of SNPs that were seen in only one of the sub-groups. The probability that a SNP occurring $k > 1$ times in an overall sample of n individuals would be found entirely within a given subset of m individuals is $B(n,k)/B(m,k)$, where $B(x,y)$ is the binomial coefficient $x!/(x-y)!y!$. In
20 this fashion, the probability that each individual SNP would be confined to a particular ethnic subgroup within the sample was calculated and these probabilities were summed to obtain the number of SNPs expected to be confined to the group within the sample. The fact that a SNP is found only within one group in the sample does not necessarily imply that it is private to that group within the general
25 population, owing to the small sample size, but it can be used as an indication of substructure. The number of SNPs with $k > 1$ confined to the, African-Americans, African Pygmies, Caucasians, and Asians was 17, 17, 12, and 9, as compared to expectations of 3.02, 1.34, 8.62, and 1.81. Not surprisingly, the greatest excess was seen for SNPs found in the African-American and African samples. The presence of
30 population substructure implies that construction of a comprehensive SNP database should employ a diverse set of DNA samples.

The results of this survey provide a fundamental description of sequence variation in the coding regions of human genes. These data indicate that two copies of a gene chosen from the human population will differ by roughly one base in 2 kb, corresponding to somewhat less than one heterozygous base within the coding region of a typical gene. In general, there are only a handful of such cSNPs per gene that exhibit allele frequencies of at least a few percent. Accounting for both the different rate and frequency of non-synonymous SNPs, only about 40% of these observed changes will alter the encoded amino acid. The action of purifying selection during human evolution is evident from the comparatively lower rate of non-synonymous cSNPs, and especially of those that create a non-conservative change. It is clear that non-synonymous cSNPs not only occur less often, but also have lower minor allele frequencies: 60% of non-synonymous cSNPs, the class likely to have the most dramatic effects on proteins, display a minor allele frequency below 5%.

The relative rarity of cSNPs has important implications for efforts to produce large catalogues of human variants. It has been proposed that most human SNPs could be found by performing shotgun sequencing on a handful of individuals (J. L. Weber and E. W. Myers, *Genome Research*, 7:401 (1997); J. C. Venter *et. al.*, *Science*, 280:1540 (1998)). Although such a project will surely identify many SNPs, results described herein suggest that the small sample size will likely fail to identify the vast majority of cSNPs likely to have the most important biological consequences, owing to their lower average allele frequencies. A comprehensive collection of the common, non-conservative cSNPs may require surveying 50-100 chromosomes. Because coding sequence represents only about 3% of the genome, it may prove inefficient to obtain such deep coverage of cSNPs by shotgun sequencing of genomic DNA. Instead, it may be more efficient to perform shotgun sequencing on cDNA libraries from multiple individuals or to amplify genes from multiple individuals, as done here.

Interestingly, a similar rate of polymorphism in coding and non-coding DNA was found. Furthermore, the observed rate of nucleotide diversity at four-fold degenerate sites was nearly twice that in adjacent non-coding regions, and over twice that at non-degenerate sites (Figure 4). Similar results have been reported for

Drosophila (E. N. Moriyama and J. R. Powell., *Mol. Biol. Evol.*, 13:261 (1996)) and for a smaller human data set by Li and Sadler (R. M. Harding *et. al.*, *Am. J. Hum. Genet.*, 60:772 (1997); D. A. Nickerson *et. al.*, *Nature Genetics*, 19:233 (1998); W. H. Li and L.A. Sadler, *Genetics* 129:513 (1991)), who observed over three times
5 the nucleotide diversity at four-fold degenerate sites ($\theta = 11 \times 10^{-4}$), as compared to that in both untranslated regions and non-degenerate sites ($\theta = 3 \times 10^{-4}$). These observations suggest that non-coding DNA adjacent to coding regions may be functionally constrained to a surprising degree.

SNPs can be used to search for genes underlying complex traits in two
10 distinct ways: linkage disequilibrium (LD) studies and association studies (E. S. Lander, N.J. Schork, *Science*, 265:2037 (1994)). Genome-wide LD studies involve using a dense collection of SNPs as markers to search for an ancestral haplotype carrying a disease-susceptibility allele. Such studies cannot be undertaken without the availability of an extremely dense SNP map and their potential for success
15 depends sensitively on many population genetic assumptions. Association studies are more straightforward because they directly test the hypothesis that a specific SNP increases disease risk. They make few assumptions, and require only the availability of a suitable database of appropriate SNPs. In the near term, focusing on cSNPs is likely to be most productive inasmuch as the class is easily recognized (in
20 contrast to regulatory polymorphisms) and is likely to contain a significant proportion of the disease-susceptibility alleles.

The present invention relates to a gene which comprises a single nucleotide polymorphism (SNP) at a specific location. The gene which includes the SNP has at least two alleles, referred to herein as the reference allele and the variant allele. The
25 reference allele (prototypical or wild type allele) has been designated arbitrarily and typically corresponds to the nucleotide sequence of the gene which has been deposited with GenBank or TIGR under a given Accession number. The variant allele differs from the reference allele by one at least one nucleotide at the site(s) identified in Figures 5A-5Q. The present invention also relates to variant
30 alleles of the described genes and to complements of the variant alleles. The invention further relates to portions of the variant alleles and portions of complements of the variant alleles which comprise (encompass) the site of the SNP

and are at least 5 nucleotides in length. Portions can be, for example, 5-10, 5-15, 10-20, 5-25, 10-30, 10-50 or 10-100 bases long. For example, a portion of a variant allele which is 5 nucleotides in length includes the single nucleotide polymorphism (the nucleotide which differs from the reference allele at that site) and four
5 additional nucleotides which flank the site in the variant allele. These nucleotides can be on one or both sides of the polymorphism. Polymorphisms which are the subject of this invention are defined in Figures 5A-5QQQQQQQ with respect to the reference sequence deposited in GenBank under the Accession number indicated. For example, the invention relates to a portion of a gene (e.g., AADC) having a
10 partial nucleotide sequence as shown in Figures 5A-5QQQQQQQ comprising a single nucleotide polymorphism at a specific position. The reference nucleotide for AADC is shown in column 3 and the variant nucleotide is shown in column 4 of Figures 5A-5QQQQQQQ. The nucleotide sequences of the invention can be double- or single-stranded.

15 The invention further provides allele-specific oligonucleotides that hybridize to a gene comprising a single nucleotide polymorphism or to the complement of the gene. These oligonucleotides can be probes or primers.

The invention further provides a method of analyzing a nucleic acid from an individual. The method determines which base is present at any one of the
20 polymorphic sites shown in Figures 5A-5QQQQQQQ. Optionally, a set of bases occupying a set of the polymorphic sites shown in Figures 5A-5QQQQQQQ is determined. This type of analysis can be performed on a number of individuals, who are tested for the presence of a disease phenotype. The presence or absence of disease phenotype is then correlated with a base or set of bases present at the
25 polymorphic site or sites in the individuals tested.

An oligonucleotide of this invention can be DNA or RNA, and single- or double-stranded. Oligonucleotides can be naturally occurring or synthetic, but are typically prepared by synthetic means. Preferred oligonucleotides of the invention include segments of DNA, or their complements, which include any one of the
30 polymorphic sites shown in Figures 5A-5QQQQQQQ. The segments can be between 5 and 250 bases, and, in specific embodiments, are between 5-10, 5-20, 10-20, 10-50, 20-50 or 10-100 bases. The polymorphic site can occur within any

Hybridization probes are oligonucleotides which bind in a base-specific manner to a complementary strand of nucleic acid. Such probes include peptide nucleic acids, as described in Nielsen *et al.*, *Science* 254, 1497-1500 (1991). Probes can be any length suitable for specific hybridization to the target nucleic acid sequence. The most appropriate length of the probe may vary depending upon the hybridization method in which it is being used; for example, particular lengths may be more appropriate for use in microfabricated arrays, while other lengths may be more suitable for use in classical hybridization methods. Suitable probes and primers can range from about 5 nucleotides to about 30 nucleotides in length. For example, probes and primers can be 5, 6, 8, 10, 12, 14, 16, 18, 20, 22, 24, 25, 26, 28 or 30 nucleotides in length. The probe or primer preferably contains at least one polymorphic site occupied by any of the possible variant nucleotides. The nucleotide sequence can correspond to the coding sequence of the allele or to the complement of the coding sequence of the allele.

As used herein, the term "primer" refers to a single-stranded oligonucleotide which acts as a point of initiation of template-directed DNA synthesis under appropriate conditions (*e.g.*, in the presence of four different nucleoside triphosphates and an agent for polymerization, such as, DNA or RNA polymerase or reverse transcriptase) in an appropriate buffer and at a suitable temperature. The appropriate length of a primer depends on the intended use of the primer, but typically ranges from 15 to 30 nucleotides. Short primer molecules generally require cooler temperatures to form sufficiently stable hybrid complexes with the template. A primer need not reflect the exact sequence of the template, but must be sufficiently complementary to hybridize with a template. The term primer site refers to the area of the target DNA to which a primer hybridizes. The term primer pair refers to a set of primers including a 5' (upstream) primer that hybridizes with the 5'

end of the DNA sequence to be amplified and a 3' (downstream) primer that hybridizes with the complement of the 3' end of the sequence to be amplified.

As used herein, linkage describes the tendency of genes, alleles, loci or genetic markers to be inherited together as a result of their location on the same chromosome. It can be measured by percent recombination between the two genes,
5 alleles, loci or genetic markers.

As used herein, polymorphism refers to the occurrence of two or more genetically determined alternative sequences or alleles in a population. A polymorphic marker or site is the locus at which divergence occurs. Preferred
10 markers have at least two alleles, each occurring at frequency of greater than 1%, and more preferably greater than 10% or 20% of a selected population. A polymorphic locus may be as small as one base pair. Polymorphic markers include restriction fragment length polymorphisms, variable number of tandem repeats (VNTR's), hypervariable regions, minisatellites, dinucleotide repeats, trinucleotide
15 repeats, tetranucleotide repeats, simple sequence repeats, and insertion elements such as Alu. The first identified allelic form is arbitrarily designated as the reference form and other allelic forms are designated as alternative or variant alleles. The allelic form occurring most frequently in a selected population is sometimes referred to as the wildtype form. Diploid organisms may be homozygous or heterozygous
20 for allelic forms. A diallelic or biallelic polymorphism has two forms. A triallelic polymorphism has three forms.

By altering amino acid sequence, SNPs may alter the function of the encoded proteins. The discovery of the SNP facilitates biochemical analysis of the variants and the development of assays to characterize the variants and to screen for
25 pharmaceutical that would interact directly with on or another form of the protein. SNPs (including silent SNPs) may also alter the regulation of the gene at the transcriptional or post-transcriptional level. SNPs (including silent SNPs) also enable the development of specific DNA, RNA, or protein-based diagnostics that detect the presence or absence of the polymorphism in particular conditions.

30 A single nucleotide polymorphism occurs at a polymorphic site occupied by a single nucleotide, which is the site of variation between allelic sequences. The site

is usually preceded by and followed by highly conserved sequences of the allele (e.g., sequences that vary in less than 1/100 or 1/1000 members of the populations).

A single nucleotide polymorphism usually arises due to substitution of one nucleotide for another at the polymorphic site. A transition is the replacement of
5 one purine by another purine or one pyrimidine by another pyrimidine. A transversion is the replacement of a purine by a pyrimidine or vice versa. Single nucleotide polymorphisms can also arise from a deletion of a nucleotide or an insertion of a nucleotide relative to a reference allele. Typically the polymorphic site is occupied by a base other than the reference base. For example, where the
10 reference allele contains the base "T" at the polymorphic site, the altered allele can contain a "C", "G" or "A" at the polymorphic site.

Hybridizations are usually performed under stringent conditions, for example, at a salt concentration of no more than 1 M and a temperature of at least 25°C. For example, conditions of 5X SSPE (750 mM NaCl, 50 mM NaPhosphate,
15 5 mM EDTA, pH 7.4) and a temperature of 25-30°C, or equivalent conditions, are suitable for allele-specific probe hybridizations. Equivalent conditions can be determined by varying one or more of the parameters given as an example, as known in the art, while maintaining a similar degree of identity or similarity between the target nucleotide sequence and the primer or probe used.

20 The term "isolated" is used herein to indicate that the material in question exists in a physical milieu distinct from that in which it occurs in nature. For example, an isolated nucleic acid of the invention may be substantially isolated with respect to the complex cellular milieu in which it naturally occurs. In some instances, the isolated material will form part of a composition (for example, a crude
25 extract containing other substances), buffer system or reagent mix. In other circumstance, the material may be purified to essential homogeneity, for example as determined by PAGE or column chromatography such as HPLC. Preferably, an isolated nucleic acid comprises at least about 50, 80 or 90 percent (on a molar basis) of all macromolecular species present.

I. Analysis of Polymorphisms

A. Preparation of Samples

Polymorphisms are detected in a target nucleic acid from an individual being analyzed. For assay of genomic DNA, virtually any biological sample (other than
5 pure red blood cells) is suitable. For example, convenient tissue samples include whole blood, semen, saliva, tears, urine, fecal material, sweat, buccal, skin and hair. For assay of cDNA or mRNA, the tissue sample must be obtained from an organ in which the target nucleic acid is expressed. For example, if the target nucleic acid is a cytochrome P450, the liver is a suitable source.

10 Many of the methods described below require amplification of DNA from target samples. This can be accomplished by e.g., PCR. *See generally PCR Technology: Principles and Applications for DNA Amplification* (ed. H.A. Erlich, Freeman Press, NY, NY, 1992); *PCR Protocols: A Guide to Methods and Applications* (eds. Innis, *et al.*, Academic Press, San Diego, CA, 1990); Mattila *et al.*, *Nucleic Acids Res.* 19, 4967 (1991); Eckert *et al.*, *PCR Methods and Applications* 1, 17 (1991); *PCR* (eds. McPherson *et al.*, IRL Press, Oxford); and
15 U.S. Patent 4,683,202.

Other suitable amplification methods include the ligase chain reaction (LCR) (see Wu and Wallace, *Genomics* 4, 560 (1989), Landegren *et al.*, *Science* 241, 1077
20 (1988), transcription amplification (Kwoh *et al.*, *Proc. Natl. Acad. Sci. USA* 86, 1173 (1989)), and self-sustained sequence replication (Guatelli *et al.*, *Proc. Nat. Acad. Sci. USA*, 87, 1874 (1990)) and nucleic acid based sequence amplification (NASBA). The latter two amplification methods involve isothermal reactions based on isothermal transcription, which produce both single stranded RNA (ssRNA) and
25 double stranded DNA (dsDNA) as the amplification products in a ratio of about 30 or 100 to 1, respectively.

B. Detection of Polymorphisms in Target DNA

There are two distinct types of analysis of target DNA for detecting polymorphisms. The first type of analysis, sometimes referred to as de novo
30 characterization, is carried out to identify polymorphic sites not previously characterized (i.e., to identify new polymorphisms). This analysis compares target

sequences in different individuals to identify points of variation, i.e., polymorphic sites. By analyzing groups of individuals representing the greatest ethnic diversity among humans and greatest breed and species variety in plants and animals, patterns characteristic of the most common alleles/haplotypes of the locus can be identified, and the frequencies of such alleles/haplotypes in the population can be determined. Additional allelic frequencies can be determined for subpopulations characterized by criteria such as geography, race, or gender. The de novo identification of polymorphisms of the invention is described in the Examples section. The second type of analysis determines which form(s) of a characterized (known) polymorphism are present in individuals under test. There are a variety of suitable procedures, which are discussed in turn.

1. Allele-Specific Probes

The design and use of allele-specific probes for analyzing polymorphisms is described by e.g., Saiki *et al.*, *Nature* 324, 163-166 (1986); Dattagupta, EP 235,726, Saiki, WO 89/11548. Allele-specific probes can be designed that hybridize to a segment of target DNA from one individual but do not hybridize to the corresponding segment from another individual due to the presence of different polymorphic forms in the respective segments from the two individuals. Hybridization conditions should be sufficiently stringent that there is a significant difference in hybridization intensity between alleles, and preferably an essentially binary response, whereby a probe hybridizes to only one of the alleles. Some probes are designed to hybridize to a segment of target DNA such that the polymorphic site aligns with a central position (e.g., in a 15-mer at the 7 position; in a 16-mer, at either the 8 or 9 position) of the probe. This design of probe achieves good discrimination in hybridization between different allelic forms.

Allele-specific probes are often used in pairs, one member of a pair showing a perfect match to a reference form of a target sequence and the other member showing a perfect match to a variant form. Several pairs of probes can then be immobilized on the same support for simultaneous analysis of multiple polymorphisms within the same target sequence.

2. Tiling Arrays

The polymorphisms can also be identified by hybridization to nucleic acid arrays, some examples of which are described in WO 95/11995. One form of such arrays is described in the Examples section in connection with de novo identification
5 of polymorphisms. The same array or a different array can be used for analysis of characterized polymorphisms. WO 95/11995 also describes subarrays that are optimized for detection of a variant form of a precharacterized polymorphism. Such a subarray contains probes designed to be complementary to a second reference sequence, which is an allelic variant of the first reference sequence. The second
10 group of probes is designed by the same principles as described in the Examples, except that the probes exhibit complementarity to the second reference sequence. The inclusion of a second group (or further groups) can be particularly useful for analyzing short subsequences of the primary reference sequence in which multiple mutations are expected to occur within a short distance commensurate with the
15 length of the probes (e.g., two or more mutations within 9 to 21 bases).

3. Allele-Specific Primers

An allele-specific primer hybridizes to a site on target DNA overlapping a polymorphism and only primes amplification of an allelic form to which the primer exhibits perfect complementarity. See Gibbs, *Nucleic Acid Res.* 17, 2427-2448
20 (1989). This primer is used in conjunction with a second primer which hybridizes at a distal site. Amplification proceeds from the two primers, resulting in a detectable product which indicates the particular allelic form is present. A control is usually performed with a second pair of primers, one of which shows a single base mismatch at the polymorphic site and the other of which exhibits perfect
25 complementarity to a distal site. The single-base mismatch prevents amplification and no detectable product is formed. The method works best when the mismatch is included in the 3'-most position of the oligonucleotide aligned with the polymorphism because this position is most destabilizing to elongation from the primer (see, e.g., WO 93/22456).

4. Direct-Sequencing

The direct analysis of the sequence of polymorphisms of the present invention can be accomplished using either the dideoxy chain termination method or the Maxam Gilbert method (see Sambrook *et al.*, *Molecular Cloning, A Laboratory Manual* (2nd Ed., CSHP, New York 1989); Zyskind *et al.*, *Recombinant DNA Laboratory Manual*, (Acad. Press, 1988)).

5. Denaturing Gradient Gel Electrophoresis

Amplification products generated using the polymerase chain reaction can be analyzed by the use of denaturing gradient gel electrophoresis. Different alleles can be identified based on the different sequence-dependent melting properties and electrophoretic migration of DNA in solution. Erlich, ed., *PCR Technology, Principles and Applications for DNA Amplification*, (W.H. Freeman and Co, New York, 1992), Chapter 7.

6. Single-Strand Conformation Polymorphism Analysis

Alleles of target sequences can be differentiated using single-strand conformation polymorphism analysis, which identifies base differences by alteration in electrophoretic migration of single stranded PCR products, as described in Orita *et al.*, *Proc. Nat. Acad. Sci.* 86, 2766-2770 (1989). Amplified PCR products can be generated as described above, and heated or otherwise denatured, to form single stranded amplification products. Single-stranded nucleic acids may refold or form secondary structures which are partially dependent on the base sequence. The different electrophoretic mobilities of single-stranded amplification products can be related to base-sequence differences between alleles of target sequences.

II. Methods of Use

After determining polymorphic form(s) present in an individual at one or more polymorphic sites, this information can be used in a number of methods.

A. Forensics

Determination of which polymorphic forms occupy a set of polymorphic sites in an individual identifies a set of polymorphic forms that distinguishes the individual. *See generally* National Research Council, *The Evaluation of Forensic*
5 *DNA Evidence* (Eds. Pollard *et al.*, National Academy Press, DC, 1996). The more sites that are analyzed, the lower the probability that the set of polymorphic forms in one individual is the same as that in an unrelated individual. Preferably, if multiple sites are analyzed, the sites are unlinked. Thus, polymorphisms of the invention are often used in conjunction with polymorphisms in distal genes. Preferred
10 polymorphisms for use in forensics are biallelic because the population frequencies of two polymorphic forms can usually be determined with greater accuracy than those of multiple polymorphic forms at multi-allelic loci.

The capacity to identify a distinguishing or unique set of forensic markers in an individual is useful for forensic analysis. For example, one can determine
15 whether a blood sample from a suspect matches a blood or other tissue sample from a crime scene by determining whether the set of polymorphic forms occupying selected polymorphic sites is the same in the suspect and the sample. If the set of polymorphic markers does not match between a suspect and a sample, it can be concluded (barring experimental error) that the suspect was not the source of the
20 sample. If the set of markers does match, one can conclude that the DNA from the suspect is consistent with that found at the crime scene. If frequencies of the polymorphic forms at the loci tested have been determined (e.g., by analysis of a suitable population of individuals), one can perform a statistical analysis to determine the probability that a match of suspect and crime scene sample would
25 occur by chance.

$p(ID)$ is the probability that two random individuals have the same polymorphic or allelic form at a given polymorphic site. In biallelic loci, four genotypes are possible: AA, AB, BA, and BB. If alleles A and B occur in a haploid genome of the organism with frequencies x and y , the probability of each genotype
30 in a diploid organism is (see WO 95/12607):

Homozygote: $p(AA) = x^2$

Homozygote: $p(BB) = y^2 = (1-x)^2$

Single Heterozygote: $p(AB) = p(BA) = xy = x(1-x)$

Both Heterozygotes: $p(AB+BA) = 2xy = 2x(1-x)$

The probability of identity at one locus (i.e., the probability that two individuals, picked at random from a population will have identical polymorphic forms at a given locus) is given by the equation:

$$p(ID) = (x^2)^2 + (2xy)^2 + (y^2)^2.$$

These calculations can be extended for any number of polymorphic forms at a given locus. For example, the probability of identity $p(ID)$ for a 3-allele system where the alleles have the frequencies in the population of x , y and z , respectively, is equal to the sum of the squares of the genotype frequencies:

$$p(ID) = x^4 + (2xy)^2 + (2yz)^2 + (2xz)^2 + z^4 + y^4$$

In a locus of n alleles, the appropriate binomial expansion is used to calculate $p(ID)$ and $p(exc)$.

The cumulative probability of identity (cum $p(ID)$) for each of multiple unlinked loci is determined by multiplying the probabilities provided by each locus.

$$\text{cum } p(ID) = p(ID1)p(ID2)p(ID3)... p(IDn)$$

The cumulative probability of non-identity for n loci (i.e., the probability that two random individuals will be different at 1 or more loci) is given by the equation:

$$\text{cum } p(\text{nonID}) = 1 - \text{cum } p(ID).$$

If several polymorphic loci are tested, the cumulative probability of non-identity for random individuals becomes very high (e.g., one billion to one). Such probabilities can be taken into account together with other evidence in determining the guilt or innocence of the suspect.

B. Paternity Testing

The object of paternity testing is usually to determine whether a male is the father of a child. In most cases, the mother of the child is known and thus, the mother's contribution to the child's genotype can be traced. Paternity testing investigates whether the part of the child's genotype not attributable to the mother is consistent with that of the putative father. Paternity testing can be performed by analyzing sets of polymorphisms in the putative father and the child.

If the set of polymorphisms in the child attributable to the father does not match the set of polymorphisms of the putative father, it can be concluded, barring experimental error, that the putative father is not the real father. If the set of polymorphisms in the child attributable to the father does match the set of polymorphisms of the putative father, a statistical calculation can be performed to determine the probability of coincidental match.

The probability of parentage exclusion (representing the probability that a random male will have a polymorphic form at a given polymorphic site that makes him incompatible as the father) is given by the equation (see WO 95/12607):

$$p(\text{exc}) = xy(1-xy)$$

where x and y are the population frequencies of alleles A and B of a biallelic polymorphic site.

(At a triallelic site $p(\text{exc}) = xy(1-xy) + yz(1-yz) + xz(1-xz) + 3xyz(1-xyz)$), where x, y and z are the respective population frequencies of alleles A, B and C).

The probability of non-exclusion is

$$p(\text{non-exc}) = 1 - p(\text{exc})$$

The cumulative probability of non-exclusion (representing the value obtained when n loci are used) is thus:

$$\text{cum } p(\text{non-exc}) = p(\text{non-exc1})p(\text{non-exc2})p(\text{non-exc3})\dots p(\text{non-excn})$$

The cumulative probability of exclusion for n loci (representing the probability that a random male will be excluded)

$$\text{cum } p(\text{exc}) = 1 - \text{cum } p(\text{non-exc}).$$

If several polymorphic loci are included in the analysis, the cumulative probability of exclusion of a random male is very high. This probability can be taken into account in assessing the liability of a putative father whose polymorphic marker set matches the child's polymorphic marker set attributable to his/her father.

C. Correlation of Polymorphisms with Phenotypic Traits

The polymorphisms of the invention may contribute to the phenotype of an organism in different ways. Some polymorphisms occur within a protein coding sequence and contribute to phenotype by affecting protein structure. The effect may be neutral, beneficial or detrimental, or both beneficial and detrimental, depending

on the circumstances. For example, a heterozygous sickle cell mutation confers resistance to malaria, but a homozygous sickle cell mutation is usually lethal. Other polymorphisms occur in noncoding regions but may exert phenotypic effects indirectly via influence on replication, transcription, and translation. A single
5 polymorphism may affect more than one phenotypic trait. Likewise, a single phenotypic trait may be affected by polymorphisms in different genes. Further, some polymorphisms predispose an individual to a distinct mutation that is causally related to a certain phenotype.

Phenotypic traits include diseases that have known but hitherto unmapped
10 genetic components. Phenotypic traits also include symptoms of, or susceptibility to, multifactorial diseases of which a component is or may be genetic, such as autoimmune diseases, inflammation, cancer, diseases of the nervous system, and infection by pathogenic microorganisms. Some examples of diseases which can be treated or diagnosed as described herein include, but are not limited to,
15 bradyarrhythmias, tachyarrhythmias, heart failure, such as congestive heart failure, congenital heart disease, rheumatic fever, valvular heart disease, cardiomyopathies, myocarditides, pericardial diseases, cardiac tumors, cardiac manifestations of systemic diseases, and traumatic cardiac injury. Other disorders include atherosclerosis, acute myocardial infarction, ischemic heart disease, hypertensive
20 vascular disease, disorders of the aorta, vascular diseases of the extremities, vessel wall disorders, such as various forms of thrombocytopenia, von Willebrand's disease and drug-induced platelet dysfunction, and homeostatic disorders relating to vessel disease and associated bleeding. Also suitable are thrombotic thrombocytopenic purpura, hemolytic-uremic syndrome, Henoch-Schönlein purpura, capillary fragility,
25 vascular purpura, metabolic and inflammatory disorders, such as those induced by rickettsiae and certain drugs, such as sulfonamides, aortic aneurysm, aortic dissection, aortic occlusion, aortitis, atherosclerosis, coronary artery disease, angina, myocardial infarction, thrombosis, hemostatic and coagulation disorders, hypertension and hypotension. Other disorders include transplant accelerated
30 vascular restenosis following balloon angioplasty, Raynaud's disease and acrocyanosis.

Additional disorders include, but are not limited to, disorders of neurodegeneration characterized by astrocyte hypertrophy including gliosis, Pick's disease, aceroplasminemia, portal-systemic encephalopathy, frontal lobe dementia and inherited and acquired ataxias, neurodegenerative diseases of other etiology including progressive supranuclear palsy, primary progressive aphasia, cortical basal degeneration, Alzheimer's disease, Huntington's disease, and Parkinson's disease, retinitis pigmentosa and amyotrophic lateral sclerosis. Other disorders include epilepsy, stroke, defects of neural migration and differentiation, including Miller-Dieker lissencephaly syndrome, and cancer of the brain including astrocytomas and gliomas, as well as psychological disorders such as schizophrenia.

Phenotypic traits also include characteristics such as longevity, appearance (e.g., baldness, obesity), strength, speed, endurance, fertility, and susceptibility or receptivity to particular drugs or therapeutic treatments.

The correlation of one or more polymorphisms with phenotypic traits can be facilitated by knowledge of the gene product of the wild type (reference) gene. The genes in which cSNPs of the present invention have been identified are genes which have been previously sequenced and characterized in one of their allelic forms.

Correlation is performed for a population of individuals who have been tested for the presence or absence of a phenotypic trait of interest and for polymorphic markers sets. To perform such analysis, the presence or absence of a set of polymorphisms (i.e. a polymorphic set) is determined for a set of the individuals, some of whom exhibit a particular trait, and some of which exhibit lack of the trait. The alleles of each polymorphism of the set are then reviewed to determine whether the presence or absence of a particular allele is associated with the trait of interest. Correlation can be performed by standard statistical methods such as a χ^2 -squared test and statistically significant correlations between polymorphic form(s) and phenotypic characteristics are noted. For example, it might be found that the presence of allele A1 at polymorphism A correlates with heart disease. As a further example, it might be found that the combined presence of allele A1 at polymorphism A and allele B1 at polymorphism B correlates with increased milk production of a farm animal.

Such correlations can be exploited in several ways. In the case of a strong correlation between a set of one or more polymorphic forms and a disease for which treatment is available, detection of the polymorphic form set in a human or animal patient may justify immediate administration of treatment, or at least the institution of regular monitoring of the patient. Detection of a polymorphic form correlated with serious disease in a couple contemplating a family may also be valuable to the couple in their reproductive decisions. For example, the female partner might elect to undergo in vitro fertilization to avoid the possibility of transmitting such a polymorphism from her husband to her offspring. In the case of a weaker, but still statistically significant correlation between a polymorphic set and human disease, immediate therapeutic intervention or monitoring may not be justified. Nevertheless, the patient can be motivated to begin simple life-style changes (e.g., diet, exercise) that can be accomplished at little cost to the patient but confer potential benefits in reducing the risk of conditions to which the patient may have increased susceptibility by virtue of variant alleles. Identification of a polymorphic set in a patient correlated with enhanced receptiveness to one of several treatment regimes for a disease indicates that this treatment regime should be followed.

For animals and plants, correlations between characteristics and phenotype are useful for breeding for desired characteristics. For example, Beitz *et al.*, US 5,292,639 discuss use of bovine mitochondrial polymorphisms in a breeding program to improve milk production in cows. To evaluate the effect of mtDNA D-loop sequence polymorphism on milk production, each cow was assigned a value of 1 if variant or 0 if wildtype with respect to a prototypical mitochondrial DNA sequence at each of 17 locations considered. Each production trait was analyzed individually with the following animal model:

$$Y_{ijkpn} = \mu + YS_i + P_j + X_k + \beta_1 + \dots \beta_{17} + PE_n + a_n + e_p$$

where Y_{ijkpn} is the milk, fat, fat percentage, SNF, SNF percentage, energy concentration, or lactation energy record; μ is an overall mean; YS_i is the effect common to all cows calving in year-season; X_k is the effect common to cows in either the high or average selection line; β_1 to β_{17} are the binomial regressions of production record on mtDNA D-loop sequence polymorphisms; PE_n is permanent environmental effect common to all records of cow n ; a_n is effect of animal n and is

composed of the additive genetic contribution of sire and dam breeding values and a Mendelian sampling effect; and e_p is a random residual. It was found that eleven of seventeen polymorphisms tested influenced at least one production trait. Bovines having the best polymorphic forms for milk production at these eleven loci are used
5 as parents for breeding the next generation of the herd.

D. Genetic Mapping of Phenotypic Traits

The previous section concerns identifying correlations between phenotypic traits and polymorphisms that directly or indirectly contribute to those traits. The present section describes identification of a physical linkage between a genetic locus
10 associated with a trait of interest and polymorphic markers that are not associated with the trait, but are in physical proximity with the genetic locus responsible for the trait and co-segregate with it. Such analysis is useful for mapping a genetic locus associated with a phenotypic trait to a chromosomal position, and thereby cloning gene(s) responsible for the trait. See Lander *et al.*, *Proc. Natl. Acad. Sci. (USA)* 83,
15 7353-7357 (1986); Lander *et al.*, *Proc. Natl. Acad. Sci. (USA)* 84, 2363-2367 (1987); Donis-Keller *et al.*, *Cell* 51, 319-337 (1987); Lander *et al.*, *Genetics* 121, 185-199 (1989)). Genes localized by linkage can be cloned by a process known as directional cloning. See Wainwright, *Med. J. Australia* 159, 170-174 (1993); Collins, *Nature Genetics* 1, 3-6 (1992).

20 Linkage studies are typically performed on members of a family. Available members of the family are characterized for the presence or absence of a phenotypic trait and for a set of polymorphic markers. The distribution of polymorphic markers in an informative meiosis is then analyzed to determine which polymorphic markers co-segregate with a phenotypic trait. See, e.g., Kerem *et al.*, *Science* 245, 1073-1080
25 (1989); Monaco *et al.*, *Nature* 316, 842 (1985); Yamoka *et al.*, *Neurology* 40, 222-226 (1990); Rossiter *et al.*, *FASEB Journal* 5, 21-27 (1991).

Linkage is analyzed by calculation of LOD (log of the odds) values. A lod value is the relative likelihood of obtaining observed segregation data for a marker and a genetic locus when the two are located at a recombination fraction θ , versus
30 the situation in which the two are not linked, and thus segregating independently (Thompson & Thompson, *Genetics in Medicine* (5th ed, W.B. Saunders Company,

Philadelphia, 1991); Strachan, "Mapping the human genome" in *The Human Genome* (BIOS Scientific Publishers Ltd, Oxford), Chapter 4). A series of likelihood ratios are calculated at various recombination fractions (θ), ranging from $\theta = 0.0$ (coincident loci) to $\theta = 0.50$ (unlinked). Thus, the likelihood at a given value of θ is: probability of data if loci linked at θ to probability of data if loci
5 unlinked. The computed likelihoods are usually expressed as the \log_{10} of this ratio (i.e., a lod score). For example, a lod score of 3 indicates 1000:1 odds against an apparent observed linkage being a coincidence. The use of logarithms allows data collected from different families to be combined by simple addition. Computer
10 programs are available for the calculation of lod scores for differing values of θ (e.g., LIPED, MLINK (Lathrop, *Proc. Nat. Acad. Sci. (USA)* 81, 3443-3446 (1984)). For any particular lod score, a recombination fraction may be determined from mathematical tables. See Smith *et al.*, *Mathematical tables for research workers in human genetics* (Churchill, London, 1961); Smith, *Ann. Hum. Genet.* 32, 127-150
15 (1968). The value of θ at which the lod score is the highest is considered to be the best estimate of the recombination fraction.

Positive lod score values suggest that the two loci are linked, whereas negative values suggest that linkage is less likely (at that value of θ) than the possibility that the two loci are unlinked. By convention, a combined lod score of
20 +3 or greater (equivalent to greater than 1000:1 odds in favor of linkage) is considered definitive evidence that two loci are linked. Similarly, by convention, a negative lod score of -2 or less is taken as definitive evidence against linkage of the two loci being compared. Negative linkage data are useful in excluding a chromosome or a segment thereof from consideration. The search focuses on the
25 remaining non-excluded chromosomal locations.

III. Modified Polypeptides and Gene Sequences

The invention further provides variant forms of nucleic acids and corresponding proteins. The nucleic acids comprise one of the sequences described in Figures 5A-5QQQQQQQ, column 11, in which the polymorphic position is
30 occupied by one of the alternative bases for that position. Some nucleic acids encode full-length variant forms of proteins. Similarly, variant proteins have the

prototypical amino acid sequences encoded by nucleic acid sequences shown in Figures 5A-5QQQQQQQ, column 11, (read so as to be in-frame with the full-length coding sequence of which it is a component) except at an amino acid encoded by a codon including one of the polymorphic positions shown in Figures 5A-
5 5QQQQQQQ. That position is occupied by the amino acid coded by the corresponding codon in any of the alternative forms shown in Figures 5A-5QQQQQQQ.

Variant genes can be expressed in an expression vector in which a variant gene is operably linked to a native or other promoter. Usually, the promoter is a
10 eukaryotic promoter for expression in a mammalian cell. The transcription regulation sequences typically include a heterologous promoter and optionally an enhancer which is recognized by the host. The selection of an appropriate promoter, for example trp, lac, phage promoters, glycolytic enzyme promoters and tRNA promoters, depends on the host selected. Commercially available expression vectors
15 can be used. Vectors can include host-recognized replication systems, amplifiable genes, selectable markers, host sequences useful for insertion into the host genome, and the like.

The means of introducing the expression construct into a host cell varies depending upon the particular construction and the target host. Suitable means
20 include fusion, conjugation, transfection, transduction, electroporation or injection, as described in Sambrook, *supra*. A wide variety of host cells can be employed for expression of the variant gene, both prokaryotic and eukaryotic. Suitable host cells include bacteria such as *E. coli*, yeast, filamentous fungi, insect cells, mammalian cells, typically immortalized, *e.g.*, mouse, CHO, human and monkey cell lines and
25 derivatives thereof. Preferred host cells are able to process the variant gene product to produce an appropriate mature polypeptide. Processing includes glycosylation, ubiquitination, disulfide bond formation, general post-translational modification, and the like.

The protein may be isolated by conventional means of protein biochemistry
30 and purification to obtain a substantially pure product, *i.e.*, 80, 95 or 99% free of cell component contaminants, as described in Jacoby, *Methods in Enzymology* Volume 104, Academic Press, New York (1984); Scopes, *Protein Purification, Principles*

and Practice, 2nd Edition, Springer-Verlag, New York (1987); and Deutscher (ed), *Guide to Protein Purification, Methods in Enzymology*, Vol. 182 (1990). If the protein is secreted, it can be isolated from the supernatant in which the host cell is grown. If not secreted, the protein can be isolated from a lysate of the host cells.

- 5 The invention further provides transgenic nonhuman animals capable of expressing an exogenous variant gene and/or having one or both alleles of an endogenous variant gene inactivated. Expression of an exogenous variant gene is usually achieved by operably linking the gene to a promoter and optionally an enhancer, and microinjecting the construct into a zygote. *See Hogan et al.*,
10 "Manipulating the Mouse Embryo, A Laboratory Manual," Cold Spring Harbor Laboratory. Inactivation of endogenous variant genes can be achieved by forming a transgene in which a cloned variant gene is inactivated by insertion of a positive selection marker. *See Capecchi, Science* 244, 1288-1292 (1989). The transgene is then introduced into an embryonic stem cell, where it undergoes homologous
15 recombination with an endogenous variant gene. Mice and other rodents are preferred animals. Such animals provide useful drug screening systems.

 In addition to substantially full-length polypeptides expressed by variant genes, the present invention includes biologically active fragments of the polypeptides, or analogs thereof, including organic molecules which simulate the
20 interactions of the peptides. Biologically active fragments include any portion of the full-length polypeptide which confers a biological function on the variant gene product, including ligand binding, and antibody binding. Ligand binding includes binding by nucleic acids, proteins or polypeptides, small biologically active molecules, or large cellular structures.

- 25 Polyclonal and/or monoclonal antibodies that specifically bind to variant gene products but not to corresponding prototypical gene products are also provided. Antibodies can be made by injecting mice or other animals with the variant gene product or synthetic peptide fragments thereof. Monoclonal antibodies are screened as are described, for example, in Harlow & Lane, *Antibodies, A Laboratory Manual*,
30 Cold Spring Harbor Press, New York (1988); Goding, *Monoclonal antibodies, Principles and Practice* (2d ed.) Academic Press, New York (1986). Monoclonal antibodies are tested for specific immunoreactivity with a variant gene product and

lack of immunoreactivity to the corresponding prototypical gene product. These antibodies are useful in diagnostic assays for detection of the variant form, or as an active ingredient in a pharmaceutical composition.

IV. Kits

5 The invention further provides kits comprising at least one allele-specific oligonucleotide as described above. Often, the kits contain one or more pairs of allele-specific oligonucleotides hybridizing to different forms of a polymorphism. In some kits, the allele-specific oligonucleotides are provided immobilized to a substrate. For example, the same substrate can comprise allele-specific
10 oligonucleotide probes for detecting at least 10, 100 or all of the polymorphisms shown in Figures 5A-5Q. Optional additional components of the kit include, for example, restriction enzymes, reverse-transcriptase or polymerase, the substrate nucleoside triphosphates, means used to label (for example, an avidin-enzyme conjugate and enzyme substrate and chromogen if the label is biotin), and
15 the appropriate buffers for reverse transcription, PCR, or hybridization reactions. Usually, the kit also contains instructions for carrying out the methods.

From the foregoing, it is apparent that the invention includes a number of general uses that can be expressed concisely as follows. The invention provides for the use of any of the nucleic acid segments described above in the diagnosis or
20 monitoring of diseases, such as coronary artery disease, diabetes, coagulation disorders, lipid metabolism disorders, energy metabolism disorders, diseases of the blood, blood vessels and cardiovascular system, and infection by microorganisms, as well as psychological disorders (e.g., bipolar disorder, schizophrenia). The invention further provides for the use of any of the nucleic acid segments in the
25 manufacture of a medicament for the treatment or prophylaxis of such diseases. The invention further provides for the use of any of the DNA segments as a pharmaceutical.

While this invention has been particularly shown and described with references to preferred embodiments thereof, it will be understood by those skilled
30 in the art that various changes in form and details may be made therein without

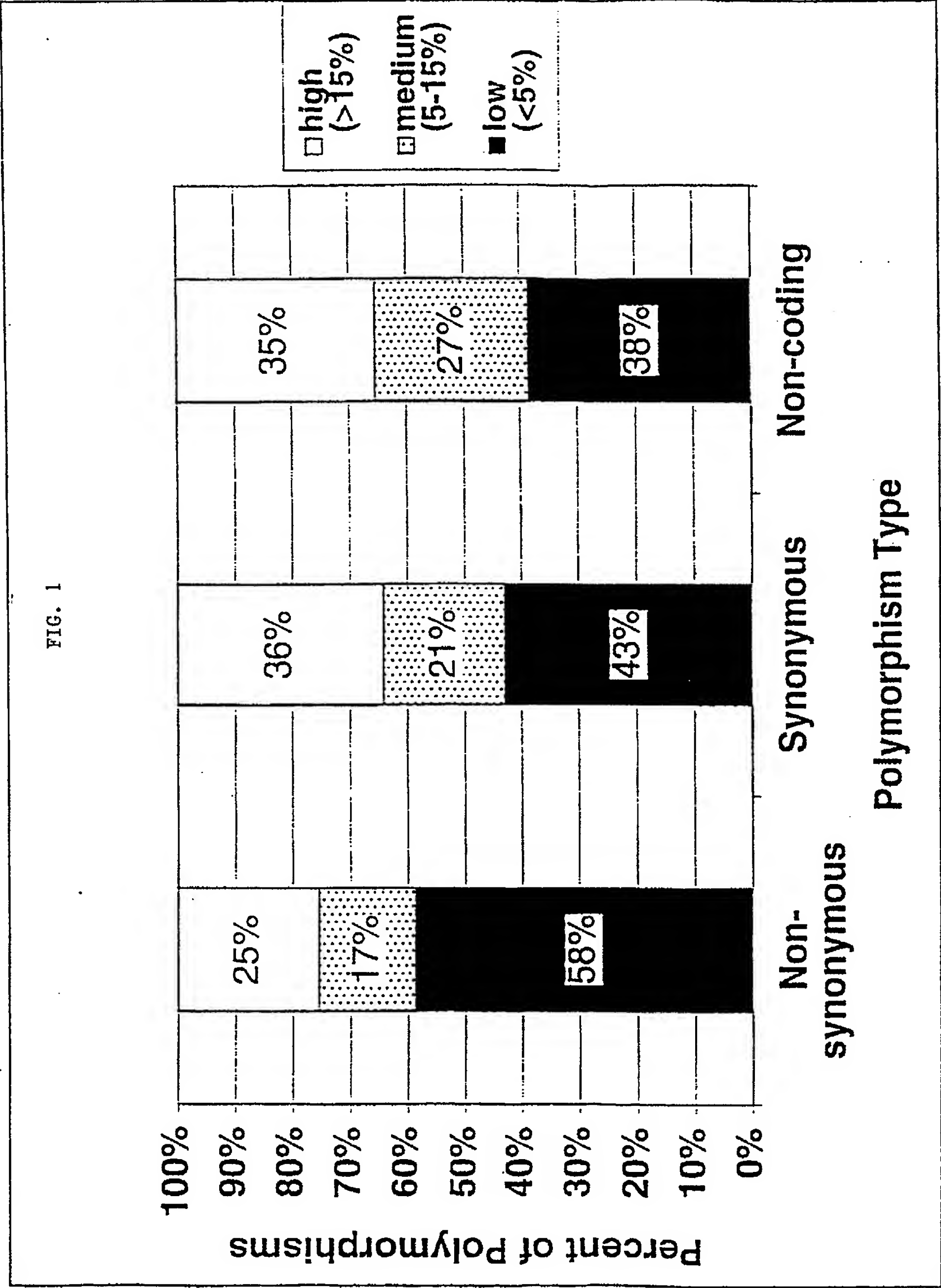
departing from the spirit and scope of the invention as defined by the appended claims.

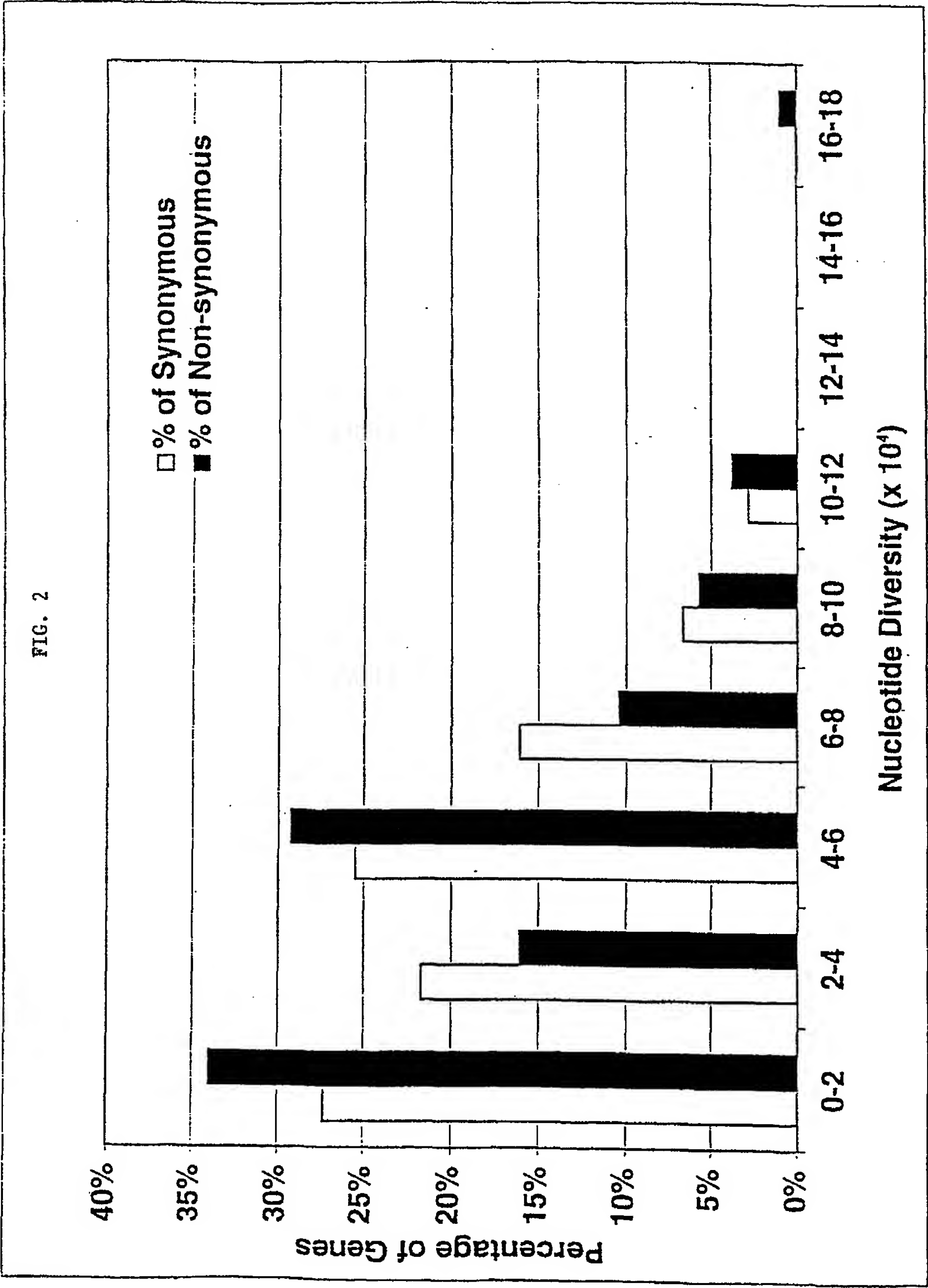
CLAIMS

What is claimed is:

1. A nucleic acid molecule selected from the group consisting of the genes listed in Figures 5A-5Q, wherein said nucleic acid molecule is at least 5 nucleotides in length and comprises a polymorphic site identified in Figures 5A-5Q, wherein a nucleotide at the polymorphic site is different from a nucleotide at the polymorphic site in a corresponding reference allele.
2. A nucleic acid molecule according to Claim 1, wherein said nucleic acid molecule is at least 10 nucleotides in length.
3. A nucleic acid molecule according to Claim 1, wherein said nucleic acid molecule is at least 20 nucleotides in length.
4. A nucleic acid molecule according to Claim 1, wherein the nucleotide at the polymorphic site is the variant nucleotide for the gene listed in Figures 5A-5Q.
5. An allele-specific oligonucleotide that hybridizes to a portion of a gene selected from the group consisting of the genes listed in Figures 5A-5Q, wherein said portion is at least 5 nucleotides in length and comprises a polymorphic site identified in Figures 5A-5Q, wherein a nucleotide at the polymorphic site is different from a nucleotide at the polymorphic site in a corresponding reference allele.
6. An allele-specific oligonucleotide according to Claim 5 that is a probe.
7. An allele-specific oligonucleotide according to Claim 5, wherein a central position of the probe aligns with the polymorphic site of the portion.

8. An allele-specific oligonucleotide according to Claim 5 that is a primer.
9. An allele-specific oligonucleotide according to Claim 8, wherein the 3' end of the primer aligns with the polymorphic site of the portion.
10. An isolated gene product encoded by a nucleic acid molecule according to
5 Claim 1.
11. A method of analyzing a nucleic acid sample, comprising obtaining the nucleic acid from an individual sample; and determining a base occupying any one of the polymorphic sites shown in Figures 5A-5QQQQQQQ.
12. A method according to Claim 11, wherein the nucleic acid sample is obtained
10 from a plurality of individuals, and a base occupying one of the polymorphic positions is determined in each of the individuals, and the method further comprising testing each individual for the presence of a disease phenotype, and correlating the presence of the disease phenotype with the base.





Gene	coding bp screened	No. Synonymous polymorphisms	No. Non- synonymous polymorphisms	Non-coding bp screened	No. Non-coding polymorphisms
AADC	1229	0	2	311	0
ADORA2	332	0	1	75	0
AHC	1413	0	0	63	1
ANX3	929	2	4	725	6
APOD	570	1	3	383	1
AR	2759	3	1	300	0
AT3	1357	3	0	121	0
BDNF	744	0	1	212	0
CD36	1209	1	1	252	0
CETP	1397	4	4	299	0
CGA	349	1	0	235	0
CLanalog	1461	3	2	12	0
CNTF	603	0	1	154	0
COMT	783	2	1	241	1
CRH	51	0	0	745	3
CYP11A	1556	1	1	547	0
CYP11B1	1410	7	7	496	9
CYP11B2	1512	7	8	906	4
CYP17	1395	3	0	36	0
CYP21	1488	5	11	542	7
DBH	1266	0	2	49	0
DRD1	1341	1	0	81	0
DRD2	1032	2	0	1379	3
DRD3	719	0	1	145	0
DRD5	1408	2	1	34	0
F10	1369	3	2	416	1
F11	1878	7	4	1312	2
F13A1	2199	3	6	948	4
F13B	1952	4	6	2339	4
F2	1740	3	2	292	0
F2R	1202	2	1	13	0
F3	875	0	1	92	0
F5	6564	13	16	1542	8
F7	1262	4	2	1209	2
F9	1364	0	1	1062	2
FGA	1935	2	2	490	0
FGB	1476	7	3	1057	0
FGG	1252	0	2	1392	2
FSH	355	1	1	44	0
FSHR	1683	1	3	0	0
GABRB1	1425	5	0	804	2
GAP43	675	1	1	79	0
GH1	644	0	1	426	5
GHR	1765	1	6	391	1
GNRHR	237	0	1	513	0
GP1BA	1881	2	2	48	0
GP1BB	1238	0	0	73	0
GP5	1683	0	0	52	0
GP9	534	1	0	143	0
GRF	224	0	0	239	0
GRIN1	1681	1	0	553	0
GRL	2334	4	3	4028	5
HCF2	1500	3	3	64	1

FIG. 3A

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Gene	coding bp screened	No. Synonymous polymorphisms	No. Non- synonymous polymorphisms	Non-coding bp screened	No. Non-coding polymorphisms
HMGCR	1724	0	1	12	1
HSD3B1	1122	3	2	653	1
HSD3B2	1122	1	1	723	2
HTR1A	1272	1	0	1189	1
HTR1D	1134	1	1	46	0
HTR1DB	1173	2	0	85	1
HTR1E	1098	1	1	70	0
HTR1EL	1101	1	0	46	0
HTR2A	1398	2	3	1709	9
HTR2C	1245	0	1	138	0
HTR5A	1062	2	0	34	0
HTR6	437	1	0	34	0
HTR7	1279	0	0	138	0
IGF1	630	0	0	7250	8
IGF2	546	0	0	610	1
ITGA2B	2833	4	3	707	0
ITGB3	2131	4	3	163	0
KLK2	297	0	1	279	2
LCAT	1289	1	2	90	0
LDLR	2101	7	3	38	0
LIPC	1471	4	3	754	4
LPL	409	1	1	48	0
MAOA	1032	1	0	69	0
MAOB	980	1	0	135	0
MPL	1748	1	2	903	1
NGFB	726	1	1	1186	5
NOS1	127	0	0	56	0
NT3	774	1	0	150	0
NTRK1	1961	5	2	1106	0
PACE	1500	2	0	1095	4
PAI1	1171	1	2	911	1
PAI2	1248	5	4	915	5
PC1	1881	1	3	456	1
PCI	1221	5	5	576	4
POMC	132	0	0	520	0
PRL	633	1	1	180	1
PROC	1334	3	0	114	0
PROS1	1868	1	0	557	0
PTAFR	1029	0	2	13	0
PTH	348	1	0	230	2
PTHLH	634	0	0	2342	13
SELP	2096	5	8	14	0
SHBG	1209	1	3	494	1
SLC6A1	1388	2	0	547	2
SLC6A3	1496	6	1	205	0
SLC6A4	1623	1	2	824	1
TBXA2R	1006	1	0	12	0
TBXAS1	1605	1	6	1411	1
TFPI	806	0	1	139	0
TH	965	1	1	104	0
THBD	1728	0	0	26	0
THPO	1049	0	0	632	2
VLDLR	2391	3	1	850	2
ALL GENES	135823	203	192	59552	150

FIG. 3B

Polymorphism rates for different classes of sites. Nucleotide diversity and heterozygosity (π) are expressed x 10⁴.

Polymorphism Type	bp screened	No. polys	Adjusted for frequency of sites*			
			Frequency (SNP/bp)	$\hat{\theta}$	π	π
Non-coding	59,552	150	1/397	4.93 ± 1.24	5.05 ± 2.40	
Coding	135,823	395	1/344	5.47 ± 1.32	5.07 ± 2.40	
synonymous		203	1/669	2.81 ± 0.68	2.98 ± 1.42	9.84 ± 2.38 10.43 ± 4.97
non-synonymous		192	1/707	2.66 ± 0.64	2.06 ± 0.98	3.73 ± 0.90 2.89 ± 1.37
conservative		122	1/1113	1.69 ± 0.41	1.44 ± 0.68	4.94 ± 1.19 4.21 ± 1.99
non-conservative		70	1/1940	0.97 ± 0.23	0.63 ± 0.30	2.61 ± 0.63 1.70 ± 0.81
four-fold degenerate sites	21,645	111	1/195	9.64 ± 2.32	9.26 ± 4.40	
two-fold degenerate sites	34,294	125	1/274	6.85 ± 1.65	5.33 ± 2.53	
non-degenerate sites	79,659	157	1/507	3.70 ± 0.89	2.52 ± 1.19	
Total	195,375	545	1/357	5.31 ± 1.28	5.01 ± 2.38	

* The number of synonymous sites was calculated as the sum of four-fold degenerate sites and half the number of two-fold degenerate sites; the number of non-synonymous sites is the sum of the non-degenerate sites and half the two-fold degenerate sites. The number of conservative and non-conservative sites is estimated as the proportion of non-synonymous sites at which a nucleotide substitution would create a conservative or non-conservative substitution, calculated as in footnote 21.

FIG. 4

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')
AADCd4	AADC	C	T	P	L	cds	GEI048	CACACACCTGT ACAAATCCAA	CTTACAAGAA AGGAATCAGGC
AADCd5	AADC	A	G	M	V	cds	GEI048	CACACACCTGT ACAAATCCAA	CTTACAAGAA AGGAATCAGGC
AADCd6	AADC	G	A	V	M	cds	GEI094	CACTGAATCAT TTTCTTTCTGC	ACACACTTACC CCAGGC
AADCd7	AADC	C	T	D	D	cds	GEI263	CCCTTGTTACT GCTGACCCC	CACCTCTCCCC CTTCTC
AADCu1	AADC	C	T	R	W	cds	GEI094	CACTGAATCAT TTTCTTTCTGC	ACACACTTACC CCAGGC
AADCu2	AADC	A	G	E	G	cds	GEI094	CACTGAATCAT TTTCTTTCTGC	ACACACTTACC CCAGGC
AADCu3	AADC	A	T	I	I	cds	GEI004	TCCATCTGGG ACTCAC	GTGCACCTPACC TCCACTC
ADORA2 _{u1}	ADORA ₂	C	T	A	V	cds	GEI141	ATGACCGTGA GCTGGC	AAGCCTGGCA CCAACA
ADORA2 _{u2}	ADORA ₂	C	G	P	P	cds	GEI141	ATGACCGTGA GCTGGC	AAGCCTGGCA CCAACA

FIG. 5A

[illegible]

[illegible]

FIG. 5E

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
ANX3u10	ANX3	T	G	L	R	cds	GE447	AAAAGTATTTT CACATTTTCC C	TGATGACTTGG TCAAACCC	AAAAGTATTTTCACTTTTCCCTTggtttttttaggagc[t/g]gaaagatgacttga agggtgatctctctgcccactttgagcatctcatggtgcccctagtgactcc[a/g]ccagcagctttt gatgcaaaagcagctaaagaaatcccatgaaggtatgagccccccacaagccatttctgcccagggt TTGACCAAGTCATCA
ANX3u11	ANX3	A	G	P	P	cds	GE447	AAAAGTATTTT CACATTTTCC C	TGATGACTTGG TCAAACCC	AAAAGTATTTTCACTTTTCCCTTggtttttttaggagctgaaagatgacttgaagg tgatctctctgcccactttgagcatctcatggtgcccctagtgactcc[a/g]ccagcagctttt gatgcaaaagcagctaaagaaatcccatgaaggtatgagccccccacaagccatttctgcccagggt TTGACCAAGTCATCA
ANX3u12	ANX3	T	G	L	R	cds	GE441	AAAAGAAATA ATTGTCCTCT AATATC	TGTAAGAACAA CATCAACACA AAGG	AAAAGAAATAATTGTCCTCTAATATCattctctgtgaatagattctctataaaagctggtgag aacagatggggcagcagtgatgaagacaatctcatgagatcc[t/g]gtgtttaaaggagcttctctc aatataaactaaggtacaaactcaccattacaatCCTTGTGTGTATGTTGTTTACA
ANX3u13	ANX3	A	G	T	T	cds	GE460	ATTCAAAATG CTCAACTGC	CTGAGTTAAAG CAAGTGCAA	ATTCAAAATGCTCAACTGCgttgctttaaataattttgtggtgcttcttttttagc[a/g]tttg atgaatacagaataatcagccaaagagacattgtgacagcagataaaaggagatttatctgggcat tttgaaagacttactgttggccataggttaagacttcgaggtcgtggttaaaactaagttactTTGCAC TGCTTTAACTCAG
ANX3u14	ANX3	T	A	I	N	cds	GE460	ATTCAAAATG CTCAACTGC	CTGAGTTAAAG CAAGTGCAA	ATTCAAAATGCTCAACTGCgttgctttaaataattttgtggtgcttcttttttagcattttaga atcacagaaata[t/a]cagccaaagagacattgtgacagcagataaaaggagatttatctgggcat tttgaaagacttactgttggccataggttaagacttcgaggtcgtggttaaaactaagttactTTGCAC TGCTTTAACTCAG
ANX3u2	ANX3	C	A	S	R	cds	GE425	CTTTTAGGGG GCGGA	AGCAACATTGG CTAAATATGTG	CTTTTAGGGGCGGGAacaaacgaagatgccttgatlgaaactcttaactaccaggacaag[c/a] laggcaaatgaagatatctctcaagcctattatacaggtgtcttattttctgttaccctcacc actgttcaCACATATTTAGCCAAATGTTGCT
ANX3u3	ANX3	C	T	P	L	cds	GE433	TTACTTACTAT AGATTAAACCA ATTTC	TTTCCCAAGGG AATTAAAGG	TTACTTACTATAGATTAAACCAATTTCTattctcgtgaagctgaacattatttctgttttttaga gttaattgtgtgggaacacgc[t/g]ggccttttagccgaagactgcacatgcagccttgaagg ttggtctggaaagttcatgtgcatcttagcgtCCCTTAATTCCTTGGGAAA
ANX3u4	ANX3	A	T	E	V	cds	GE444	TCTCTATGTT CTTTGTGACCA AT	GACTTACTTTA ATTGCTGAATA TAGG	TCTCTATGTTGTTGTGACCAATgacatttgggtgtggaacacctgcattcttaacagggtat tggaactgatgagttactctgaacccaataatggtgtccagatcag[a/t]aattgaccttttg gacattcgaacagagttcgaagacatttatggctatttccCTATATTTCAGCAATTAAAGTAAGTC
ANX3u5	ANX3	C	T	-	-	noncoding	GE439	TGATTCAATTA TGGTCTCCCAT T	GAAGTAAGGTG GAGCTGTTGG	TGATTCAATTTATGTTCTCCCAATtatttatactgtatttgttttctcatgtatttatt[c/t]ttt gcagtcggataacttctgagagactatgaaatcacactcttaaaaaactctgtggtgagatgactgaa ccaaagagataatctccaaaggctccacgatggggttttCCCAACAGCTCCACCTTACTTC
ANX3u6	ANX3	G	T	-	-	noncoding	GE448	CTTTTGTTC GACAAATGTTG AG	TGATCTCTTAC TGCCTGTCA	CTTTTGTTCAGACAAATGTTGAGCataaatatttgagtaaaaaactttt[g/t]tttctatttag gaactgatgagaaatgctcatcagcattcttgactgagaggtcaaatgcacagggcagctgatt gttaagggaataatcaagcagcatatggaaaggtaagggtcacattacaatGACAGGAGTAAGAGAT CA
ANX3u7	ANX3	A	T	E	D	cds	GE425	CTTTTAGGGG GCGGA	AGCAACATTGG CTAAATATGTG	CTTTTAGGGGCGGGAacaaacga[a/t]gatgcttgattgaaatcttaactaccaggacaag caggcaaatgaagatatctctcaagcctattatacaggtgtcttattttctgttaccctcacc actgttcaCACATATTTAGCCAAATGTTGCT
ANX3u8	ANX3	T	G	-	-	noncoding	GE443	GATGTCATTT GAACCAATG	TGTAACCTGCC TGATTTGCTTC	GATGTCATTTTGAACCAATGgacttttcaagatt[t/g]cctttaggttggaacccaggagaca gtaagagattatccagacttttagcccatcagtggtgctgaagctatttcagaaagcaatcagagg aatgggtgagtgattttacaattcttcttctaatgttGAAGCAAAATCAGGCAAGTTACA

FIG. 5D

[illegible]

FIG. 5F

[illegible]

FIG. 5G

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
ARU8	AR	T	C	S	S	cds	GE568	CTCCTTGTCAA CCCTGTTT	GGGCATGAGCT GGGGTG	CTCCTTGTCAAACCTGTTTCTCCTCCTTATTGTTCCCTACAGATGCGGAGAGAGCTGCAATCAG TTCACTTTTGACCTGCTAATCAAGTCAACATGTTGAGCGTGGAGCTTCCGGAATGATGGCAGA GATCATCTCTGTGCAAGTGCCCAAGATCCTTCT/CTGGGAAGTCAAGCCCATCTATTCCAC ACCGAGTGAAGCATGGAAACCTATTCCCAACCCAGCTCATGCC	243
BDNFu1	BDNF	G	A	V	M	cds	GE1184	AAGCCCTAACC AGTTTCTG	GTTCCTTCT GGTCATGGA	AAGCCCTAACCAGTTTCTGCTTGTCTCCTACAGTTCCACAGGTGAGAGAGT GATGACCATCTTCTCCTTACTATGTTTCTCCTACACTTGGTTCATGAGAGGTGCCCCATGA AAGAGCAACATCCGAGGACCAAGTGGCTGGCTACCAAGGTGTCGGAACCATGGGACTCTG GAGAGCGTGAATGGGCCCCAAGGCTTCAAGAGGCTTGACATCATGGTGACACTTCGAACA CGTGATAGAGAGCTGTGGATGAGGACCAAGTTCGGCCCAATGAGGAACCAATAGGACG CAGACTTGTACAGTCCAGGCTGATGCTCAGTAGTCAAGTGGCTTGGAGCCTCCTCTCTCTT CTGCTGGAGGGAACAAAATACCTAGATGCTGCAACATGCTCAGTGGTCCGGGCGCCACTC TGCCCTGCCCGGAGGAGTGGAGCTGTGTGACAGTATTAGTGGTGGTAAAGGCGGCA ACAAAAGACTCAGTGGACATGTCGGGGGACGGTCAAGTCCCTGAAAAGGTCCTGTATCA AAGGGCCCACTGAAGCAATCTCTACGAGACCAAGTGAATCCCATGGGTACACAAAAGAGG CTGAGGGGATAGACAAAAGGCTTGGAACTCCAGTCCGGAACCTACAGTCTGAGTGGG CCCTTACCATGATAGCAAAAAGAGAAATGGCTGGCGATTCAAGGALT/G)AGACCTCTCTG TGATGTACATTGACCAATTAAGGAGGAGATAGTGGATTATGTTGATAGATTAGATTAT GAGACAAAATATCTATTGTATATATACATAACAGGGTAAATATTCAAGTTAAGAAAAAATA ATTTATGAAGTCAATGATAAATGAAGTTTATACAGTACAGTGGTTCACAACTATTATGG ACATGTCCATGACCAAGAGGAAAC	100
BDNFu2	BDNF	T	G	I	R	cds	GE1184	AAGCCCTAACC AGTTTCTG	GTTCCTTCT GGTCATGGA	AAGCCCTAACCAGTTTCTGCTTGTCTCCTACAGTTCCACAGGTGAGAGAGT GATGACCATCTTCTCCTTACTATGTTTCTCCTACACTTGGTTCATGAGAGGTGCCCCATGA AAGAGCAACATCCGAGGACCAAGTGGCTGGCTACCAAGGTGTCGGAACCATGGGACTCTG GAGAGCGTGAATGGGCCCCAAGGCTTCAAGAGGCTTGACATCATGGTGACACTTCGAACA CGTGATAGAGAGCTGTGGATGAGGACCAAGTTCGGCCCAATGAGGAACCAATAGGACG CAGACTTGTACAGTCCAGGCTGATGCTCAGTAGTCAAGTGGCTTGGAGCCTCCTCTCTCTT CTGCTGGAGGGAACAAAATACCTAGATGCTGCAACATGCTCAGTGGTCCGGGCGCCACTC TGCCCTGCCCGGAGGAGTGGAGCTGTGTGACAGTATTAGTGGTGGTAAAGGCGGCA ACAAAAGACTCAGTGGACATGTCGGGGGACGGTCAAGTCCCTGAAAAGGTCCTGTATCA AAGGGCCCACTGAAGCAATCTCTACGAGACCAAGTGAATCCCATGGGTACACAAAAGAGG CTGAGGGGATAGACAAAAGGCTTGGAACTCCAGTCCGGAACCTACAGTCTGAGTGGG CCCTTACCATGATAGCAAAAAGAGAAATGGCTGGCGATTCAAGGALT/G)AGACCTCTCTG TGATGTACATTGACCAATTAAGGAGGAGATAGTGGATTATGTTGATAGATTAGATTAT GAGACAAAATATCTATTGTATATATACATAACAGGGTAAATATTCAAGTTAAGAAAAAATA ATTTATGAAGTCAATGATAAATGAAGTTTATACAGTACAGTGGTTCACAACTATTATGG ACATGTCCATGACCAAGAGGAAAC	1000

FIG. 5H

[illegible]

Fig. 5.

[illegible]

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
CYP11A 1a3	CYP11A A	G	A	E	K	cds	GE585	CTGCAGGGAAC CTCACTCTT	CACAGGGGCA ACAAGGT	CTGACGGGAACCTCACTCTTTTtttttttctctcttccccacagctgacatatacacccgaactt ctactgggaattggacagaaaggaagtgttaccacgataccctggcactctctacagactcc tgggagacagcaagatgtcttc(g/a)aggacatcaaggccaagctcacagagatgttggcagg aggggtggacacaggtgaggtggctgtagggggcACCTTGTGTGCCCCCTGTG	248
CYP11A 1a4	CYP11A A	T	A	L	Q	cds	GE556	CTACTCCCCAC CAGACG	CCAGGGATTGG AGTTGGG	CTACTCCCCACACGACGtccatgacctgacgtggcacttggatgagatggcacaacctgaag gtgcagatatgtctgcgggacagaggtcttggctgcggcgacacagggcccagggagacatggccac gatgtacagctgttccccctctcaagcagcatcaaggagacac(t/a)aaggcaaggccacac aacacccatgcccgcCCCACTTCAATCCCTGG	229
CYP11B 1a30	CYP11B BI	A	T	N	I	cds	GE570	TCCCAGCACCA AAGTCTGAG	GGCATCACCTT CTCTGGGT	TCCCAGCACCAAGTCTGAGgggtgcctccccgtccccggataggcgacaactgtatccagaaaa tctatcaggaactgcttcagccgcctcaacagtatcacacgataccacgcatctgtggcgagctctctgttg a(a/t)tgcggaactgtcccgagatgccatbaaggccaactctatggaaactcaactgcagggagcg tggacacggtcagggcggaacacagccccACCACGAGAGGGGTGATGCC	243
CYP11B 1a31	CYP11B BI	A	G	-	-	noncoding	GE577	GAATGGGCTG AATGGC	CTCCAGGGTCT CTGAGGCTG	GAATGGGCTGAATGGCGcttcaaccgatggggctgaatccagaagtgtctgtcgccccacgctg tgcagaggttctctccgatggttgatgcagtgccagggactctccccagggccttgaaagaaag gtctgcagaaacgcccggggagcctgacctggacgtccagccacgcatcttccactataccat agaaggtctgggcaac(a/g)tgggaagatccAGCCTCAGAGACCCCTGGAG	246
CYP11B 1a32	CYP11B BI	A	T	-	-	noncoding	GE617	ATGGCACTCAG GGCAAA	AGGGCTCTGGG TGTTCCTC	ATGGCACTCAGGGCAAGgcagaggtgtgcattggcagtgccctggctgccccgaaggacaca ggacttgggcacagagccgcgggtccccagggctcccaagacagtgtgcctttgaagccatgccacgc gtccaggcaacaggtggctgaggtgtctgcagatctggaggagcagggttatgaggacttgacac ctggaagtacacccagccttccaggaactggggcccatcttcaaggtaaggcctctctggccc(a /t)cgctGGGAACCCACAGAGCCCT	285
CYP11B 1a33	CYP11B BI	C	T	-	-	noncoding	GE625	GGAGGCAGCCA GGAGGC	GTGTCCCTTCC CCATAGCAC	GGAGGCAGCCAGGAGGC(c/t)cggggtgccttgtgtcagcagtgcatctccccgaagccag caacttggctctcttttggagagggctggggccttgggttggccacagccccagttctgtgccagctg aacttctccatgcctggaggtcatgttcaatccacgtccagctcatgttcatgtccacaggag cctgtctgctggaccagccccaaaggtgtggaaggagcacttggggcctgggactgcattctcc agtaagggtgaggccaggaccgggcaGTGTATGGGAAGGGACAC	307
CYP11B 1d24	CYP11B BI	G	C	-	-	noncoding	GE1231	GCAACTTTGAG GGTCTGAGAA	CCTGGGTGAG GCAGAAA	GCAACTTTGAGGGTCTGAGAAgggtgcaccagctcgatgggtcggaacagccagatggaaac cc(g/c)gctgtgcaccaggtgctgaaacactccaggtggagacactaacccaagagacata aagatgggtctacaggttcatattgaggccccagcatgttccccctctcaactcagagccatcaa ctaatacagctctctgcacccaggggtccagcctggccacacgctctctcttTCTGCTGACCCACAG G	261
CYP11B 1d25	CYP11B BI	T	C	Y	Y	cds	GE570	TCCCAGCACCA AAGTCTGAG	GGCATCACCTT CTCTGGGT	TCCCAGCACCAAGTCTGAGgggtgcctccccgtccccggataggcgacaactgtatccagaaaa tcta(t/c)caggaaactggccttcagccgcctcaacagtatcacacgcatctgtggcgagctcct gttgatgcggaaactgtccagatgccataaggccaactctatggaaactcaactgcagggagcg tggacacggtcagggcggaacacagccccACCACGAGAGGGGTGATGCC	243
CYP11B 1d26	CYP11B BI	C	G	N	K	cds	GE577	GAATGGGCTG AATGGC	CTCCAGGGTCT CTGAGGCTG	GAATGGGCTGAATGGCGcttcaaccgatggggctgaatccagaagtgtctgtcgcccaa(c/g) gctgtgcagaggttctctccgatgggtgagatggcagggacttccccagggccttccagggcctgaagaa gaagtgtctgcagaacgcccggggagcctgacctggagactccagccagcatcttccactaca cctaataaggtgtggccsacatgggaagatccAGCCTCAGAGACCCCTGGAG	246

FIG. 5L

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
CYP11B 1d27	CYP11B B1	C	A	-	-	noncoding	GE582	CTCCTGTGCAA GGTCTG	CTCCAGCAGGG GGCCAG	CTCCGTGTGCAAGGTCTGaccctgcagctgtgtctctcctgcagacgggtgtttcccttgctgatgaac gctctttgagctggctcgaaacccaaacgtgcagcagggccctgcagagagagcttgccgcggc cagccagcatcagtgaaatccccagaaaggaacacacagagctnccccctgtgctgctgctgctgct caaggagaccttg[c/a]ggtgggtgctggctgagggcctccctgtggccctgGCCCCCCCTGCTGGA G
CYP11B 1d28	CYP11B B1	C	T	-	-	noncoding	GE617	ATGGCACTCAG GGCAAA	AGGGCTCTGGG TGTTCCT	ATGGCACTCAGGGCAAAggcagaggtgtgcattggcagtgccctgggctgtccctgcaaaagggcaca ggcactggggcacagagcggccgggtccacagacagtgctgcctttgaagccatgccccagc gtccaggcaaacaggtggctgaggtgctgcagatctggaggagcaggggtatgaggacctgcac ctggaagtacacacagacctccaggaaatggggccatttccaggtaaaagccctcc[c/t]tgggc ccacgtGGGAACACCCAGAGCCCT
CYP11B 1d29	CYP11B B1	C	T	-	-	noncoding	GE625	GGAGGCAGCCA GGAGGC	GTGTCCCTTCC CCATAGCAC	GGAGGCAGCCAGGAGGC[c/t]ggggctgctctgtgtcagcagtgcatcctcccccagccag caacttggctctttttggagagcggctgggctgggttggccacagcccccaagttcttgcagcctg aaacttccctccactggctgaggtcatgttcaaatccacctccactcatgttcatgccccaggag cctgtctgctggaccagcccccaaggtgtggaagayagcaatttgaggcctgggactgcatcttcc agtaagggtgagggccagggccggcagTGCTATGGGAAAGGACAC
CYP11B 1u1	CYP11B B1	A	G	Q	R	cds	GE617	ATGGCACTCAG GGCAAA	AGGGCTCTGGG TGTTCCT	ATGGCACTCAGGGCAAAggcagaggtgtgcattggcagtgccctgggctgtccctgcacaaagggcaca gycactggggcacagagcggccgggtccacagacagtgctgcctttgaagccatgcccc[a/ g]ggtccaggcaaacaggtggctgaggtgctgcagatctggaggagcaggggtatgaggacct gcacctggaagtacacacagaccttccaggaaactggggccatttccaggtaaaagccctccctggc ccacgtGGGAACACCCAGAGCCCT
CYP11B 1u10	CYP11B B1	C	G	P	A	cds	GE582	CTCCTGTGCAA GGTCTG	CTCCAGCAGGG GGCCAG	CTCCTGTGCAAGGTCTGaccctgcagctgtgtctctcctgcagacgggtgttcccttgctgatgaac gctctttgagctggctcgaaacccaaacgtgcagcagggccctgcagagagacctggccggcgg cagccagcatcagtgaaatccccagaaaggaacacacagagctn[c/g]cccttgctgctgctgctg cccctcaaggagaccttgsggtgggtgctggctgagggcctccctgtggccctTGGCCCCCTGCTGGA G
CYP11B 1u11	CYP11B B1	T	G	-	-	noncoding	GE582	CTCCTGTGCAA GGTCTG	CTCCAGCAGGG GGCCAG	CTCCTGTGCAAGGTCTGaccctgcagctgtgtctctcctgcagacgggtgttcccttgctgatgaac gctctttgagctggctcgaaacccaaacgtgcagcagggccctgcagagagacctggccggcgg cagccagcatcagtgaaatccccagaaaggaacacacagagctnccccctgtgctg[c/t/g]gagg ccctcaaggagaccttgsggtgggtgctggctgagggcctccctgtggccctTGGCCCCCTGCTGGA G
CYP11B 1u12	CYP11B B1	C	T	L	L	cds	GE531	ACAGGAAGCCC CATCCA	AGGTTCCTCAGC TCGAGGGGT	ACAGGAAGCCCCCATCCAGctgaggacctttctatggatgccccacacctccagggtctacacctgt gggctgtgtt[c/t]tgaggcaggtgtgtaggtctcagacttggtgcttcagaactaccacatcccc gctgggtgagtgagccccacACCCCTCGAGCTGAGAACCT
CYP11B 1u13	CYP11B B1	T	C	L	L	cds	GE618	CTCCCCAGTCA TTCCCTGA	GCCCATGCTGC CCAGAC	CTCCCCAGTCAATTCCTGATccccgctctgcaccgtccgcagacatgggtgaggtgttctctcta ctctctgggtcgcaacccccgcttgttcccgagggcctgagcgtatataacccccagcgtggctag acatcaggggctccggcaggaacttctaccacgtcccttgggttggcattggccagtgacct[c t/c]gggcggcgctggcagaggcagagatgctgctgctgctgacacctgtgagcagggccccgggc tggggaggggcctgggcgggTCTGGGCAGCATGGGC
CYP11B 1u14	CYP11B B1	G	A	C	Y	cds	GE617	ATGGCACTCAG GGCAAA	AGGGCTCTGGG TGTTCCT	ATGGCACTCAGGGCAAAggcagaggtgt[g/a]catggcagtgccctggctgtccctgcacaaaggg cacaggcactgggcacagagccggccgggtccccaggacagtgctgaccttgaagccatgccc cagcgtccaggcaaacaggtggctgaggtgctgcagatctggaggagcaggggttatgaggacct gacctgggaagtacacacagaccttccaggaaactggggccatttccaggtaaaagccctccctggc ccacgtGGGAACACCCAGAGCCCT

FIG. 5M

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Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
CYP11B 1u15	CYP11 B1	G	A	R	H	cds	GE617	ATGGCACTCAG GGCAA	AGGGCTCTGGG TGTTCCC	ATGGCACTCAGGGCAAAGgagaggtgtgcatggcagtgccctggctgtccctgcaaaagggcaca ggcactgggcaagagagccggggtcccccagagacagtgctggccctttgaagccatgccccagc [g/a]tccaggcaacaggtggctgaggtgctgagctgctgagatctggagggagcaggttatgaggaact gcacctggaagtacaccagaccttccaggaaactggggcccaatttcagggraaagccctccctggc ccacgctGGGAACACCCAGAGCCCT	285
CYP11B 1u16	CYP11 B1	C	T	D	D	cds	GE536	ATGGTCCCATTT CCAGCAC	AGCAAGAACAC GCCACA	ATGGTCCCATTTCCAGCACgggctcgtgcttggccccacaggtacga[c/t]tgggagggagcagg catggtgtgtgtgctgctgcccggagacgtggagaaagctgcaacaggtggacagcctgcatcccc acaggatgagcctggagccctgggtggcctacagacaacatcgtgggcacaaaTGTGGCTGTTC TTGCT	200
CYP11B 1u17	CYP11 B1	G	A	L	L	cds	GE625	GGAGGCAGCCA GGAGGC	GTGTCCCTTCC CCATAGCAC	GGAGGCAGGCAGGAGGCCggggctgcttgtgctcagcagtgcatcctccccgaagccagcaac tt[g/a]gctcttttggagagcggctgggcttggccacagcccccaagtcttggccagcctg aacttctccatgcccctggaggtcatgttcaaatccaccgtccagctcatgttcatgcccagag cctgtctcgtggaccagcccccaaggtgtggaaggagacacttggagccctgggactgcatcttcc agtacggtgagggccaggggaccgggcaGTCATGGGGAAGGACAC	307
CYP11B 1u18	CYP11 B1	C	T	-	-	noncoding	GE570	TCCAGCACCA AAGTCTGAG	GGCATCACCT CTCTGGGT	TCCAGCACCAAGTCTGAGggctgctcc[c/t]gctccccggataggcacaactglatccag aaaatctatcaggaactggccttcagccctcaacagtaacagacagcctggtggggagactcct gttgaatgaggaactgtgcagatgcccacacagctcaagggcccaactctatggaactcactgcaggagc tggacacggtcagggccggcagccagccccACCAGAGAGGGGTGATGCC	243
CYP11B 1u19	CYP11 B1	G	A	A	T	cds	GE582	CTCTGTGCAA GGTCTG	CTCCAGCAGGG GGCCAG	CTCTGTGCAAGGTCTGacctgcagctgtgctcctgcagaggtgttcccttgcctgalgaac gctctttgagctggtcggaaaccccaacgtgcagcagccctggccagggagagcctggccgccc g/a]cagccagctcagtgaaacatccccagaggcaacacagagctnccccctgctgctgctggg ccctcaaggagaccttgctgggtgctgctggctgagggcctccctgtggccCTGGCCCCCTGTCTGGA G	261
CYP11B 1u2	CYP11 B1	A	G	E	E	cds	GE617	ATGGCACTCAG GGCAA	AGGGCTCTGGG TGTTCCC	ATGGCACTCAGGGCAAAGgagaggtgtgcatggcagtgccccggctgtccccgcaaaagggcaca ggcactgggcaagagagccggggtcccccagagacagtgctggccctttgaagccatgccccagc gtccaggaacacaggtggctgaggtgctgcagatctggagggagcaggggttatgaggaacctgcac ctggaagtacaccagaccttccagga[a/g]ctggggccccattttcaggtaaaagccctccctggc ccacgctGGGAACACCCAGAGCCCT	285
CYP11B 1u20	CYP11 B1	T	C	V	A	cds	GE531	ACAGGAAGCCC CATCCA	AGGTTCTCAGC TCGAGGGGT	ACAGGAAGCCCATCCAGctgagagccctttctatggatgccccacccctccaggctctaccctgt gggtctgtttctggagcggagtggt[t/c]gagctcagacttgggtgcttcagaaactacacatcccc gctggggtgagtgagcccccaCCTCCCTCGAGCTGAGAACCT	171
CYP11B 1u21	CYP11 B1	C	G	R	R	cds	GE618	CTCCCCAGTCA TTCCCTGA	GCCCATGCTGC CCAGAC	CTCCCCAGTCAATTCCTGAtccccgctctgcacccgtccgcagacaaatgggtgcgctgtccctta ctctctgggtcgcaacccccgcttgttccccgagccctgagcgtataaacccccagcgtggctag acatcaggggctccggcaggaacttctaccagctgccccttggctttggcatggccagtgccctt gggcccgg[c/g]ctggcagagggcagagatgctgctgctgcacccaatgtgagcagggccccgggc tggggaggggcttgggggGTCCTGGGACGATGGGC	297
CYP11B 1u22	CYP11 B1	A	T	T	S	cds	GE625	GGAGGCAGCCA GGAGGC	GTGTCCCTTCC CCATAGCAC	GGAGGCAGGCAGGAGGCCgggggctgcttgtgctcagcagtgcatcctccccgaagccagcaac ttggctcttttggagagggctgggctgtgtggccacagcccccaagtcttgcagcctgaact tccctccatgcccctggaggtcatgttcaaatccaccgtccagctcatgttcatgcccagggagcctg tctcgtgtg[a/t]ccagcccccaaggtgtggaaggagacacttggagggcctgggactgcatcttcc agtacggtgagggccaggggaccgggcaGTCATGGGGAAGGACAC	307

FIG. 5N

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
CYP11B lu23	CYP11B1	C	G	-	-	noncoding	GE577	GAATGGGCGCTG AATGGC	CTCCAGGCTCT CTGAGGCTG	GAATGGGCGCTGAATGGCGCTTCAACGATTCGCGGTGAATCCAGAACTGCTGTGCGCAACGCTG TGCAGAGTTCTCTCCGATGGTGGATGCGAGTGGCGAGGACTTCTCCAGGCGCTGAAGAAG GTGCTGCAGAACGCGCGGGAGCTGACCTGGAGTCCAGCTCCAGCCAGCATCTTCCACTACACCAT AGAAGTGTGGGCA[C/G]ATGGGAAGATCCAGGCTCAGAGACCTCGAG
CYP11B lu3	CYP11B1	G	A	L	L	cds	GE617	ATGGCACTCAG GGCAA	AGGCTCTGGG TGTTC	ATGGCACTCAGGGCAAAAGGCAGAGGTGTGCATGGAGTGCCTGGTGTCTCCGTCAAAAGGCGACA GGCACTGGGCAACGAGAGCGCGGGTCCCAAGGAGTGTGCTGCTTGAAGCCATGCCCGAGC GTCCAGGCAACAGTGGCTGAGGTGTGCAGATGTGGAGGAGAGGTTATGAGGACCTGCAC CTGGAATACACAGACTTCCAGGAAT[G/A]GGGCCATTTCCAGTAAAGCCCTCCCTGGC CCAGCTCGGAACACCCAGAGCCCT
CYP11B lu4	CYP11B1	C	A	F	L	cds	GE577	GAATGGGCGCTG AATGGC	CTCCAGGCTCT CTGAGGCTG	GAATGGGCGCTGAATGGCGCTTCAACGATTCGCGGTGAATCCAGAACTGCTGTGCGCAACGCTG TGCAGAGTTCTCTCCGATGGTGGATGCGAGTGGCGAGGACTTCTCCAGGCGCTGAAGA GAAGTGTGCTGCAGAACGCGGGAGCTGACCTGGAGTCCAGCTCCAGCCAGCATCTTCCACTACA CCATAGAAGGTGTGGGCCACATGGGAAGATCCAGGCTCAGAGACCTCGAG
CYP11B lu5	CYP11B1	A	G	K	R	cds	GE577	GAATGGGCGCTG AATGGC	CTCCAGGCTCT CTGAGGCTG	GAATGGGCGCTGAATGGCGCTTCAACGATTCGCGGTGAATCCAGAACTGCTGTGCGCAACGCTG TGCAGAGTTCTCTCCGATGGTGGATGCGAGTGGCGAGGACTTCTCCAGGCGCTGAAGA GAAGTGTGCTGCAGAACGCGGGAGCTGACCTGGAGTCCAGCTCCAGCCAGCATCTTCCACTACA CCATAGAAGGTGTGGGCCACATGGGAAGATCCAGGCTCAGAGACCTCGAG
CYP11B lu6	CYP11B1	C	T	T	I	cds	GE625	GGAGGCAGCCA GGAGGC	GTGTCCTTCC CCATAGCAC	GGAGGCAGGCAGGAGCGCGGGTGTGCTGTGCTCAGCAGTGCATCTCCCGAAGCAGCAAC TTGGTCTTTTGGAGAGCGGTGGGCTGGTGTGGCCACAGCCCAAGTCTGCGAGCTGAAC TCTCCATGCCCTGGAGTCAATGTTCAATCCACGTCACGTCATGTCATGCCAGGAGCCTG TCTGCTGGA[C/T]CAGCCCCAAGTGTGGAAAGGACACTTGGAGCCTGGAGTGCATCTTCC AGTACGGTGAAGGCCAGGACCGGCACTGTCTATGGGGAAGGACAC
CYP11B lu7	CYP11B1	G	A	A	A	cds	GE570	TCCAGCACCA AAGTCTGAG	GGCATCACCT CTCTGGGT	TCCAGCACCAAAAGTCTGAGGCTGTGCTCCGCTCCCGGTAGGAGCAACTGTATCCAGAAA TCTATCAGGAACCTGGCTTCAGCGGCTCAACAGTACACCACTGCTGGC[G/A]GAGCTCCT GTGAATGCGGAAGTGTGCGAGTGCATCAAGGCAACTCTATGGAACCTCACTGAGGAGCG TGGACAGGTCAAGGCGGCAACCGCCCCACCCAGAGAGGTGATGCC
CYP11B lu8	CYP11B1	G	A	-	-	noncoding	GE570	TCCAGCACCA AAGTCTGAG	GGCATCACCT CTCTGGGT	TCCAGCACCAAAAGTCTGAGGCTGTGCTCCGCTCCCGGTAGGAGCAACTGTATCCAGAAA TCTATCAGGAACCTGGCTTCAGCGGCTCAACAGTACACCACTGCTGGC[G/A]GAGCTCCT AATGGAAGTGTGCGAGTGCATCAAGGCAACTCTATGGAACCTCACTGAGGAGCTGGA CAGGTCAAGGCG[G/A]GCAACCAAGCCCCACCCAGAGAGGTGATGCC
CYP11B lu9	CYP11B1	A	G	N	D	cds	GE582	CTCCTGTGCAA GGTCTG	CTCCAGCAGGG GGCCAG	CTCCTGTGCAAGGTCTGACCTGCACTGTGCTCCTGCAGACGGTGTCTCCCTGTGATGAAC GCTCTTGAAGCTGGCTGGAACTT[G/G]ACGTGCAGCAGGCTTGCCTCAGGAGGCTGGCC GCCGCAAGCATCAGTGAACATCCCAAGAGGCAACCAAGCTTGTGCTGCTGGG CCCTCAAGGAGACTTGGGTGGGTGTGGTGAAGGCTCCCTGTGGGCTGTGGCCCTGTGCTGGA G
CYP11B 2d22	CYP11B2	C	T	-	-	noncoding	GE1213	GAGACTGAAG GGAGTGTG	CCACTGGGTGG TGGAGA	GAGACTGAAGGGAGTGTGGGAGGAGCCAGGAGGCGCGGTGTGCTTGTGCTCAGCAGTGA TCTCCCGCAGCAGCAACTAGTCTCTTTGGAGAGCGGTGGGCTGTGTTGGCCACAGCCCC AGTCTGCCAGGCTGAACCTTCTCATGCTTGGTGGTGTGTTCAAACTCCAGTCCAGTCAAT GTCTATGCCAGGAGCTGTCTGTGATCAGCCCCAAGGTGTGGAAAGGAGCACTTGAAGGCT GGGACTGCATCTCCAGTACGTGAGGCGGAGGAGGCGGAGTGTGCTATGGGAGGAGCAACAT GGGGGCCCAATTTCTCC[C/T]TCTCCACCAACCCAGTGG

FIG. 50

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Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
CYP11B 2d23	CYP11 B2	G	A	-	-	noncoding	GE533	CATCCAGCTGA GGACCCCTT	ACTGGGAGGG AGGTTCTC	CATCCAGCTGAGGACCCCTTct [g/a] tggatgccccacacccaggtctaccctgtgggtctg tcttttggagcagtggtgagctcagactggtgcttcaagaactaccacatcccagctgggtgag tgagccccacacccctcgagctGAGAACCTCCCTCCCTCCCT	172
CYP11B 2d24	CYP11 B2	A	G	N	S	cds	GE587	ATGCTTCCAG CACCAAGAT	GGCATCACCT CTCTGGG	ATGCTTCCAGCACCAAGATctgagggtgtccctgtccctggacaggtgacaaactgtatcca gaaatctaccaggaactggccttca [a/g] ccgccccacacactacacaggtcatcgtggcagag ctcctgttgaaggcggaactgtcactagagcccatcaaggccaaactctatggaaactcactgcagg gagcgtggacacaggtcaggccagcaaacagcccccaCCCAGAGGGTGTATGCC	248
CYP11B 2d25	CYP11 B2	A	C	R	R	cds	GE588	GAGTCTCTCTG TGCAAGGTC	CTCCAGCAGGG GGCCAG	GAGTCTCTCTGCAAGGTCagaccctgagacatggcttctgttagacaggttctcccttgcga tgacgtcttctgagctggtcggaaccccgacgtgacagatcctgcgcaggagagagcctggcc ggcgacgcagcatcagtgaaacatccccagagggcaacacccaggtgctgcctggcggggc cctcaaggagaccttg [a/c] ggtgggtgctggatgaggcctccctgtggcctGGCCCCCTGCT GGAG	264
CYP11B 2d26	CYP11 B2	G	A	-	-	noncoding	GE610	CCTGTGCTCTG CTGGGG	CAGGGTCTCTG GGGCTG	CCTGTGCTCTGCTGGGGggcctcacaaagctctgcctggcctctgtaggaaatgggacctgaatgg cgcttcaaccgattgcggctgaacccagatgtgctgtcgcccaaggccgtgcagaggttctctccc gatgtggtgagtgagtgccagggacttctccagccctgaagaagaaggtgctgcagaaacgcc ggggagcctgacccctggacgtccagccagcatcttccactacacatagaaggtgtggggccat gggggaag [g/a] tccAGCCCCAGAGACCCCTG	292
CYP11B 2u1	CYP11 B2	A	G	K	R	cds	GE610	CCTGTGCTCTG CTGGGG	CAGGGTCTCTG GGGCTG	CCTGTGCTCTGCTGGGGggcctcacaaagctctgcctggcctctgtaggaaatgggacctgaatgg cgcttcaaccgattgcggctgaacccagatgtgctgtcgcccaaggccgtgcagaggttctctccc gatgtggtgagtgagtgccagggacttctccagggccctga [a/g] gaagaaggtgctgcagaaac ggccggggagcctgacccctggacgtccagccagcatcttccactacacatagaaggtgtggg ccatgcgggaaggtcCAGCCCCAGAGACCCCTG	292
CYP11B 2u10	CYP11 B2	G	T	R	R	cds	GE588	GAGTCTCTCTG TGCAAGGTC	CTCCAGCAGGG GGCCAG	GAGTCTCTCTGCAAGGTCagaccctgagacatggcttctgttagacaggttctcccttgcga tgacgtcttctgagctggtcggaaccccgacgtgacagatcctgcgcaggagagcctggcc ggcgacgcagcatcagtgaaacatccccagagggcaacacccaggtgctgcctgtgctg [g/t] g cgccctcaaggagaccttgaggtgggtgctggatgaggcctccctgtggcctGGCCCCCTGCT GGAG	264
CYP11B 2u11	CYP11 B2	G	A	A	T	cds	GE637	TCCTGGGTGAG ATAAAGGATT T	AGGGATCTGGG TGTTCCT	TCCTGGGTGAGATAAAGGATTtgggtgaacaggtgagggagcatctggaaatggcactcaggg caaaggcagaggtgtgctgtggcagccctggctgtccctgcaaaaggccagggcactgggcaact agagcc [g/a] ctcgggcccttaggaaggtgtgctgctgttgaagccatgcccagcatccaggca acaggtggtgaggtgctgcagatctggaggagagcaggttatagacacctgcacctggagatg caccagaccttccaggaggtggggcccatcttcaggtaaaagccctccctggcctcgtGGGAAC ACCCAGATCCCT	337
CYP11B 2u12	CYP11 B2	G	A	P	P	cds	GE637	TCCTGGGTGAG ATAAAGGATT T	AGGGATCTGGG TGTTCCT	TCCTGGGTGAGATAAAGGATTtgggtgaacaggtgagggagcatctggaaatggcactcaggg caaaggcagaggtgtgctgtggcagccctggctgtccctgcaaaaggccagggcactgggcaact agagccgtcggggcccttaggaaggtgtgctg [g/a] ttcgaagccatgcccagcatccaggca acaggtggtgaggtgctgcagatctggaggagagcaggttatagacacctgcacctggagatg caccagaccttccaggaggtggggcccatcttcaggtaaaagccctccctggcctcgtGGGAAC ACCCAGATCCCT	337
CYP11B 2u13	CYP11 B2	A	G	A	A	cds	GE587	ATGCTTCCAG CACCAAGAT	GGCATCACCT CTCTGGG	ATGCTTCCAGCACCAAGATctgagggtgtccctgtccctggacaggtgacaaactgtatcca gaaatctaccaggaactggccttcaaccgcccccaacactacacaggtcatcgtggc [a/g] gag ctcctgttgaaggcggaactgtcactagaagccatcaaggccaaactctatggaaactcactgcagg gagcgtggacacaggtcaggccagcaaacagcccccaCCCAGAGGGTGTATGCC	248

FIG. 5P

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
CYP11B 2u14	CYP11B2	T	C	V	A	cds	GE533	CATCCAGCTGA GGACCCCTTT	ACTGGGAGGG AGGTTCTC	CATCCAGCTGAGGACCCCTTTTctgtggatgccccccacctccaggtctacacctgtacacctgtgggtctgtttt tggagcagtggtt/c)gagctcagacttgggtgtctcagaacctccacacatcccagctggggtgag tgagccccacacccctcgagctGAGAACCTCCCTCCCACT	172
CYP11B 2u15	CYP11B2	A	C	T	P	cds	GE1214	CCAGCTGGGGT GAGTGAG	CCAGTGTGCAG GTCCCG	CCAGCTGGGGTGAAGAGccccccacacccctcgagctgagaaacctccccctccccagtcattccctg atccctgctctgcaccgtccgcag[a/c]cattggtacaggttttctctactctccctccccagtcattccctg atgcgccttctcccgaggtgagcgtataatccccagcgtggtt/a)agacatcaggggtcc ggcaggaacttccaccacgtgaccttggcttggcatgagcagtgctcgggagcgcctggc agagggcagagatgctgctgctgcacacagtaagcaggcctgggggaggggagcctggg cagcagaggCGGACCTGCACACTGG	351
CYP11B 2u16	CYP11B2	T	A	L	Q	cds	GE1214	CCAGCTGGGGT GAGTGAG	CCAGTGTGCAG GTCCCG	CCAGCTGGGGTGAAGAGccccccacacccctcgagctgagaaacctccccctccccagtcattccctg atccctgctctgcaccgtccgcagacatgggtacaggttttctctactctccctccccagtcattccctg cgcttcttcccgaggtcgagcgtataatccccagcgtggtt/a)agacatcaggggtcc ggcaggaacttccaccacgtgaccttggcttggcatgagcagtgctcgggagcgcctggc agagggcagagatgctgctgctgcacacagtaagcaggcctgggggaggggagcctggg cagcagaggCGGACCTGCACACTGG	351
CYP11B 2u17	CYP11B2	G	A	V	M	cds	GE1214	CCAGCTGGGGT GAGTGAG	CCAGTGTGCAG GTCCCG	CCAGCTGGGGTGAAGAGccccccacacccctcgagctgagaaacctccccctccccagtcattccctg atccctgctctgcaccgtccgcagacatgggtacaggttttctctactctccctccccagtcattccctg cgcttcttcccgaggtcgagcgtataatccccagcgtggtt/a)agacatcaggggtcc ggcaggaacttccaccac[g/a]tgaccttggcttggcatgagcagtgctcgggagcgcctggc agagggcagagatgctgctgctgcacacagtaagcaggcctgggggaggggagcctggg cagcagaggCGGACCTGCACACTGG	351
CYP11B 2u18	CYP11B2	G	A	-	-	noncoding	GE652	CATGGGCTACT GACCAAGC	CAGGCTGCAGG AGGAA	CATGGGCTACTGACCAAGCagatggaacccagcctctgtctcaggtgctggaagcacttccctg tggagacactaactcagaggacataaagatggtctacagcttcatatttggagcctggcagctcc ccccctcacttccagagcgtataatagctcttgcacatgcacccaggttccacgctggccac cagcttccctctgctgacccagcagcctgtcttctctccac[g/a]tgacagcttccctga gtcaccctctgtccagcagctcctgcacaaatggaaactccccagggcctccagggactgggct tggcaggtctgtcaaatagcaaggccagcacagctggagacgacttctgtggcagggcctggc ctgtccccagccccacotggcccttctccagcagcagtgccctctggacagcttgactctac tccctccagcgtggctccaggtctctcatgagggcactgcaggggtgctgtgatttggctccctg ccttccctgctagctcactgctcctgctcctctgcctcggcagggcctctgtgcagacagt gtcagagctcattaaaggaggatccccagcaltctcagagtcacaggtTCCCTCTGTCAGCCTG	648
CYP11B 2u19	CYP11B2	G	C	-	-	noncoding	GE637	TCCCTGGGTGAG ATAAAAGGATT	AGGGATCTGGG TGTTCCC	TCCCTGGGTGAGATAAAAGGATTggtgggtggaacaggggtg[c]agggagcattggaatggcactc agggcaaaaggcagagggtgtgctggtggcagcgcctggtgtccctgcacaggcagggcactggg cactagagccgctcgggccctcaggacggtgctgctgcttggaaagcactgccccagcattccagga acaggtggtcgtgagctgtgcagatctggagggagcaggggttatgagacactgcacctggagatg caccagaccttccagaggctggggggccalttctcaggttaaaggcctccccctccccctggcctggcgaac ACCCAGATCCCT	337
CYP11B 2u2	CYP11B2	A	G	K	K	cds	GE1213	GAGGACTGAAG GGAGTGTG	CCACTGGGTGG TGGAGA	GAGGACTGAAGGAGTGTGGggggaggaagccagaggagccccgggggtgcttgtgctcagcagtgca tccctccccgcagccagcaacttagctcttcttggagaggggtggcctgggttggccacagcccc agttctgcagcgtgaacttctccatgctcctggaggtcatgtcaa[a/g]tccacgtccagc tcagtctcatgccccagggagcctgtctgctggatcagccccaaaggtgtggaaggagcactttagg gcctgggactgcacttccagtaggtgagggccagggcagctgctatggggaaggagga ccatggggggcccaattctctctctTCCACCCACCCAGTGG	364

FIG. 59

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
CYP11B 2u20	CYP11 B2	G	C	L	L	cds	GE588	GAGTCTCTCTG TGCAAGGTC	CTCCAGCAGGG GGCCAG	GAGTCTCTCTGTCAGAGGTGagaccctgcagacatcgtggtctctgtagacagcgtttcccttgctga tgacgtctcttgagctggctcggaaaccccgacgtgagcagatctctgcccagagagccttgccc gcccagccagcatcagtgaaatccccagaaagcaaccacagact[g/c]cccttgctgcggg cgccctcaaggagacacttgagggtgggtgctggtgagggcctccctgtggccctGGCCCCCTGCT GGAG
CYP11B 2u21	CYP11 B2	A	C	S	R	cds	GE552	CATGGGCTACT GACCAGGC	CAGGCTGCAGG AGGGAA	CATGGGCTACTGACCGCCagatggaaacccagcctclgtcctagggtgctgaagcacttccctgg tggagacactaactcaagagagacataaagatggtctac[a/c]gcttcattatgagccttgccac gtccctctctcacttccagagcgttaactagtctgcactgcacccaggtccagcctgg ccaaagcttcccttgctgacccagggccactgtctctccacgtgcacagcttcctga gtcacccctctgtccagccagctcctgcacaaatggaactccccagggcctccaggaactgggct tgccaggtctgtcaaatagcaaggccagggcacagctggagacgactctgtgagggcclggc ctgtccccagccccacactggcccttctccagcagcagtgccctctggacagcttgactctac tccctccagcgtggtccagctcctcatgagggccatgcaagggtgctgtgatttgcctctg ccttcctgcttagtctcaacatgtccctgtccctctgcacctggccagggcctctgtgcagacagt gtcagagtcattaaaggggatccccagcatctcagagtcacagtcacagTCCCTCCAGCCCTG
CYP11B 2u3	CYP11 B2	C	G	A	A	cds	GE1213	GAGGACTGAAG GGAGTGTC	CCACTGGGTGG TGGAGA	GAGGACTGAAGGGAGTGTGgggagcagccagggagggccgggctgcttgtgctcagcagtgca tcctcccgagcagcagcaacttagctcttttggagagcggtggcctggctggccacagcccc agttctgcagcctgaacttctccatgccttgaggtctatgttcaaatccaccgtccagctcat gttcagtcgccagggagcctgtctgctggatcagcccccaagggtgtggaaggagcactttgaggc[c /g]tgggactgcactctcagtaaggtgagggcagggagccccgggcagtgctatggggaagggaca ccatggggggcccaattctctctTCCACACCCAGTGG
CYP11B 2u4	CYP11 B2	G	A	A	A	cds	GE587	ATGCTTCCAG CACCAAGAT	GGCATCACCT CTCTGGG	ATGCTTCCAGCACCAAGATctgagggtgtccccctgctccccggagaggtgacaactgtatcca gaaaatctaccaggaactggccttcaaccgcccctcaactacacagggcatcgtggcagagctcc tgttgaaaggc[g/a]gaactgtcactagaagccatcaaggccaaactctatggaaactcactgcagg gagcgtggacacaggtcagggccagcaaccagcccccaCCAGAGAGGGTGTATGCC
CYP11B 2u5	CYP11 B2	T	C	I	T	cds	GE588	GAGTCTCTCTG TGCAAGGTC	CTCCAGCAGGG GGCCAG	GAGTCTCTCTGTCAGAGTcagaccctgcagacatggctctctgtagacagcgtttcccttgctga tgacgtctcttgagctggctcggaaaccccgacgtgcagcaga[t/c]cctgcgccagagagcct ggccggccagccagcatcagtgaacatccccagaaaggcaaccacagagctgccttgctgcgggg cgccctcaaggagacacttgagggtgggtgctggatgagggcctccctgtggccctGGCCCCCTGCT GGAG
CYP11B 2u6	CYP11 B2	G	A	G	S	cds	GE1214	CCAGCTGGGT GAGTGAG	CCAGTGTGCAG GTCCCG	CCAGCTGGGGTGTAGTGAGcccccaacccctcgagctgagaacctccctccccagtcattccctg atccctgctctgcacgctccgcagacattggtacaggttttccctcactcgtcgtggctgcgaatgc cgcttgttccccagggcctgagcgtataatccccagcgtggctgagacatcagggggtcc[g/a]gcaggaacttccaccacgtgcctttggctttggcatgcgccagtcctcggggcgccctggc agaggcagagatgtgctgctgctgcaccacgttaagcagggcctggggcgggggagacctggg cagcagagggCGGGACCTGCACACTGG
CYP11B 2u7	CYP11 B2	G	A	R	Q	cds	GE637	TCCTGGGTGAG ATAAAAGGATT T	AGGGATCTGGG TGTTCCT	TCCTGGGTGAGATAAAAGGATTggggctgaacaggttgagggagagattggaatggcactcaggg caaggcagaggtgtgcgtggcagcgccctggctgtccctgcgaaggccagcggtcgtggcact agagccgctc[g/a]ggccccctagacaggtgctgcctttgaagccatgccccagcatccaggca acaggtggctgaggtgctgcagatctggagggaggggttatgagcacctgcactggagatg caccagaccttccagagagctgggggccattttcaggttaaaagccctccctggcctgcgtGGGAAC ACCCAGATCCCT

FIG. 5B

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
CYP11B B2	CYP11B2	C	T	F	F	cds	GE610	CTGTGCTCTG CTGGGG	CAGGGTCTCTG GGGCTG	CCTGTGCTCTGTCTGGGGGGCCCTCACAAGCTCTGCCCTGGCTCTGTAGGAATGGGCCCTGAATGG CGCTCAACCGATTGGGCTGAACCAAGATGTCTGTGCCCCAAGCGCTGCAAGAGTCTCTCTCC GATGTTGATGCACTGGCCAGGACTT[C/T]TCCAGGCCCTGAAGAAGGTGCTGCGAGAAC GCCGGGGAGCCTGACCTGGAGCTCCAGCCAGCATCTCCACTACACCATAGAAGGTGTGGG CCATGGGGAAAGTCCAGCCCCAGAGACCTG
CYP11B B2u9	CYP11B2	T	C	I	T	cds	GE1213	GAGACTGAAG GGAGTGTG	CCACTGGGTGG TGGAGA	GAGACTGAAGGGAGTGTGGGAGGCGAGGAGGCGGGGCTGCTGTGCTCAGCAGTGA TCTCCCGCAGCCAGCAACTTAGCTTTTGGAGAGCGGCTGGGCTGGTGTGGCCACAGCCCC AGTCTCCGAGCCTGAATCTCTCCATGCTGGAGGTCATGTTCAATCCACCGTCCAGCTCAT GTTCTAGCCAGGAGCTGTCTGCTGGA[T/C]CAGCCCCAAGGTGGGAAGGAGCATTGAG GCCGTGGAGTGCATCTCCAGTACGGTGGAGCCAGGAGCCCGGCGAGTGTCTATGGGAAGGGACA CCATGGGGGCCCAATTTCTCTCCACCACCCAGTGG
CYP17u 1	CYP17	C	T	II	II	cds	GE626	CTTCTACTCCA CTGCTGTCTAT C	GGCACCACTT ACCATT	CTTCTACTCCACTGCTGTCTATCTTGGCTGCGGCAACCCAGCCACCATGTGGAGCTGTGGCTC TCTGTGCTTACCCTAGCTTATTTGTTTGGCCCAAGAGAGTGCCTGGTGCCTGCTGCTC AAGAGCTCCTGTCTGCTGCTGCTGGGAGCTGCTTCTCCCAAGACAGGCTATGCA TGATAAACAATCTTCAAGCTGCAAAAAATATGGCCCATCTATCGTCTGATGGGCACC AAGACTACAGTGATTGTGGGCCCAACCAAGCTGGCCAGGAGGTGCTTATTAAGAAGGGCAAGGA CTCTCTGGGGGCTCAATGGTAAGTGTGCTCC
CYP17u 2	CYP17	G	T	S	S	cds	GE626	CTTCTACTCCA CTGCTGTCTAT C	GGCACCACTT ACCATT	CTTCTACTCCACTGCTGTCTATCTTGGCTGCGGCAACCCAGCCACCATGTGGAGCTGTGGCTC TCTGTGCTTACCCTAGCTTATTTGTTTGGCCCAAGAGAGTGCCTGGTGCCTGCTGCTC AAGAGCTCCTGTCTGCTGCTGCTGGGAGCTGCTTCTCCCAAGACAGGCTATGCA TAACAATCTTCAAGCTGCAAAAAATATGGCCCATCTATC[G/T]GTTGCTGTTGGGCACC AAGACTACAGTGATTGTGGGCCCAACCAAGCTGGCCAGGAGGTGCTTATTAAGAAGGGCAAGGA CTCTCTGGGGGCTCAATGGTAAGTGTGCTCC
CYP17u 3	CYP17	G	A	P	P	cds	GE641	CCTTGCTGCA GAGCGT	GGGCACATAG GGTGGGA	CCTTGCTGCAAGCGTCTTGAATCAGCGGGGACCCAGCTCATCTCACC[G/A]TCAGTAAG CTATTGGCTCTCGGAGCAGGACCTGCTCTGTATAGTGTAGATCTCGGCCCGCCAGGAGCTCT TCTCATCATGGCTGGCTGCTGCAAGGTTCGACCTGGAGGTGCCAGATGATGGCAGCTGCC TCCGTGAAGGCATCCCAAGTGGTCTTCTGACGACTCTTCAAGTGAAGTCAAGGTGCG CCAGGCTGGAGGAAAGCCAGGCTGAGGTGAGCACTAAAGGTGTAACTCACAGCCCTGTCC ACCTATGTGGGCC
CYP17u 4	CYP17	T	A	L	Q	cds	GE626	CTTCTACTCCA CTGCTGTCTAT C	GGGCACCACTT ACCATT	CTTCTACTCCACTGCTGTCTATCTTGGCTGCGGCAACCCAGCCACCATGTGGAGCTGTGGCTC TCTGTGCTTACCCTAGCTTATTTGTTTGGCCCAAGAGAGTGCCTGGTGCCTGCTGCTC AAGAGCTCCTGTCTGCTGCTGCTGGGAGCTGCTTCTCCCAAGACAGGCTATGCA TAACAATCTTCAAGCTGCAAAAAATATGGCCCATCTATCGTCTGTTGTTGGCCCAAGA CTACAGTATTGTGGGCCCAACCAAGT[A/G]GGCCAAAGGAGGTGCTTATTAAGAAGGGCAAGGA CTCTCTGGGGGCTCAATGGTAAGTGTGCTCC
CYP17u 5	CYP17	C	T	S	F	cds	GES20	TCTCTAAAGGC AACTCTAGACA TC	CTGGCACTCAC TGATCTTC	TCTCTAAAGGCAACTCTAGACATCGCTCCCAACCAACCGTAAGGGTATCGCTTCTGCTGCTC TGGCCACACTGGCAGCTGATCGAAGGCTGGGAGTGGCCACCTTTGCCCTGTCTCAAGGATGGC GATCAGGAGCTGGGAAGATCAGTGAAGTCCAG
CYP17u 6	CYP17	G	A	G	D	cds	GES20	TCTCTAAAGGC AACTCTAGACA TC	CTGGCACTCAC TGATCTTC	TCTCTAAAGGCAACTCTAGACATCGCTCCCAACCAACCGTAAGGGTATCGCTTCTGCTGCTC G[A]CGCACTGGCAGCTGATCGAAGCTGGGAGTGGCCACCTTTGCCCTGTCTCAAGGATGGC GATCAGGAGCTGGGAAGATCAGTGAAGTCCAG

FIG. 55

[illegible]

FIG. 5F

[illegible]

FIG. 5U

[illegible]

FIG. 5V

[illegible]

FIG. 5W

[illegible]

FIG. 5X

[illegible]

FIG. 5Y

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
DRD1u5	DRD1	A	C	A	A	cds	GE1173	CTGACCCCTAT TCCCTGCTT	CTCTCCAAGGC CGCAATG	CTGACCCCTATTCCTGCTAGgaacttgaggggtgtcagagccctgatgtgctttctcttagg aagatgaggactctgaacacctctgccatggacgggactgggtggtggtgagagggacttctc tgcttgatctcctcactgctgtttctctgtgctgtcctcctgtccacgctcctctggggaacacgc tggtctgtgctgctggttctcaggttcgcacacctgggtccaagtgacaaactcttctgtcatc tcccttggtgtgtcagatctcttgggtggtggtcctgtgtcatgcccgggaagcagtggtgagat tgctggcttctggcccttgggtcctctgttaacatctgggtggcctttgacatcatgtgtctcca ctgc(a/c)tcacatcctcaacctctgtgtatcagcgtggacaggtatctgggtctatctccagccc tttccgggtatgagagaaagatgaccccccaaggcagccttcatcctgatcagtggtgcatggacct tgctgtactcatctcctcatccagtgacgtcagctcagtcagtcaggaagcaaaacccacaagcccc tctgatggaaatgccacttccctggctgagaccatagacaactgtgactccagcctcagcaggac atatgccatctcatcctctgtaataagcttttacatcctgtggccatcatgatgtgcacctaca ccaggatctacagggttgctcagaaaacaatacggcgCATTCGGGCTTGGAGAG
DRD1u6	DRD1	A	G	K	E	cds	GE1170	GGATCTACAGG ATTGCTCAGAA AC	GCAATCTCCTC TAGCTTTTGG	GGATCTACAGGATTGCTCAGAAAcaaatcgcgcgcattgcggccttgagagggcagcagctccac gccaagaattgccagacacacacacaggttaattggaaagcctgtcgaaatgtctcacaacggaaagttc tttaagatgtccttcaaaagagaaactaaagctcctgagacactctgtcgtgatatcatgggtgtgt ttgtgtgctgtggctacctttcttcacatctgaactgtatctgtgtgggtctgtgggtgag acgcagcccttctgcatgtatctcaaacacctttgacgtgtttgtgtgtttgggtgggtcaattc atccttgaccccatcatattatgccttaattgctgattttcggaaaggcattttcaacctctctag gatgtacagactttgcctgcagcaaatatgccatagacggtgagtatcaataacaatggg gccgcgagtgtttccagccatcatagccacgaggctccatctccaaggagtgcaatctctggttta cctgatccacatgtgtgggtcctctgaggaacctgaaagaggaggaagcagctggcatcgcca gaccttgagg(a/g)agctgtccacgacctatcggtcatattggacatgatgacactgacgtctc tctggagaagatccaaaccatcacacaaacggtcagaccccaacctgaaactgcagatgaatcc tgccacacatgctcatcccaaaagctTAGAGGAGATTG
DRD1u7	DRD1	A	C	R	R	cds	GE1173	CTGACCCCTAT TCCCTGCTT	CTCTCCAAGGC CGCAATG	CTGACCCCTATTCCTGCTAGgaacttgaggggtgtcagagccctgatgtgctttctcttagg aagatgaggactctgaacacctctgccatggacgggactgggtggtggtgagagggacttctc tggttgatctcctcactgctgtttcctgtcgtgctcatcctgtccacgctcctctggggaacacgc tggtctgtgctgcttatacaggttcg(a/c)cacctgcggtccaagtgacaaactcttctgt catctccttggtgtgtcagatctcttggtggcgtctctggtcatgccctggaaagcagtggtgtg agattgctggcttctggcccttgggtcctctgtaaacatgggtggcctttgacatcatgtgc tccactgcatccatcctcaacctctgtgtgatcagcgtggacaggtatctgggtctatctccagccc tttccgggtatgagagaaagatgaccccccaaggcagcctcatcctgatcagtggtggacct tgctgtactcatctcctcatccagtgacgtcagctcagtcagtcaggaagcaaaacccacaagcccc tctgatggaaatgccacttccctggctgagaccatagaaactgtgactccagcctcagcaggac atatgccatctcatcctctgtaataagcttttacatcctctggccatcatgatgtgcacctaca ccaggatctacagggttgctcagaaaacaatacggcgCATTCGGGCTTGGAGAG

FIG. 5Z

[illegible]

FIG. 5AA

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
DRD2u3	DRD2	A	G	-	-	noncoding	GEI1315	GCACGTGGGTTG GGTGT	GGTCTCCAGG ACTGAAGTT	GCACGTGGGTTGCGGTTCccagcgcctgccccggcctctggggaccagcctgacacatgcccttccccaggcggtgttcattcatctgttggtgcccctttctcatcacacatatctgaacatacaactgtgactgcaaacatccccgcctgtccctgtacagcgccttcacgtggctgggttatgtcaacagcgccgtgaacccaatcatcacacacctcaaatgtagtccgaagccttccctgaagatccccaatgtgactgtgtgctgcccgcacagcgcctgttccacacctccctgccagggc[a/g]gcagcentcaccttggaaacgtgagcnaggaaggcctgggtgga tcggcctcctcttcannncc ccggcaggccccgcagtgctgcgttggttgcctcactcaccgcgcacacctcactctgcc agggcagtgctagtggctlgygcattggtaaccagcccccttccctggggctggnccccccagctcaggggcag ctcatagagtcccccctccacctccagtcaccttcccttggcacaagaatgcagcggccttc cttgaccttcccttggggcttagggttgctgagcctgagtcagggcccccagagctgagtttcc tcttgtggggcttggcgtggagcggcgtggggagagatggacaggttcacacccctgcaaggcc cacaggaggcaagcaagctctctggcggaggagccaggAAC TTCAC TCTGGGAGACC
DRD2u4	DRD2	C	T	P	P	cds	GEI140	GCTGATGCCGTG GGAACTT	GGAA TGGGACC TTTCACA	GCTGATGCC TGGGAAC TTgtccggcttaccacagagcctctgcccctgtggtgcaggaggtgcccc ggagccccaggagctggagatggagatgctctccagcaccagccccaccgcagagaccgggtac agccccatccccaccagccaccaacagctgacttccccgacacctccccacatgggtctccacag cactcc[c/t] gacagccccgccaaaaccagagagaatgggcatgccaagaccacccccaaagatt gccaaagatctttgagatccagaccatgcccaaaggcaaacccggacctcccccaagacatgag ccgtaggaaagctctccagcagaaaggaagaaagcactcagatgctcggcattgttctcggtg agtcggccctggctgctggccacagtcgtctGTGTAAGGTCCCATTCC
DRD2u5	DRD2	G	T	-	-	noncoding	GEI165	CCACAGGAGGC AAGCAAG	GACTCGTCAAA GTTTTATTAGT TTGGT	CCACAGGAGC CAAGAGctctcttgcgagggagccaggcaacttcagtcctgggagacccatgt aaataccagactgcaggtlgyaccccagagatgccaaagccaaaaaaccttagctccctcccgcac ccgatgtggacctctacttcccaggctagtcctcgaccacacctcaccctgtacagctccccaaag tgggttccacatgctctgagaagagagccctcatcttgaagggccagggaggtctatgtggggag aggaactccttggcctlagccacacctlg/t ctgccccttgagcggcctgcaatgtatcccttct cacagcaatgctggccagcctggggcctggcaggggtcagggcctggaaactctatctgggcc tgggttaggggacatcagaggttctttgaggggactgctctgcccactctgacgcaaaaacacact tccctttctattccttctggccttctctctctctgttcccttcccttccactgccctlgcct tagaggagccccacggctaagagggtgctgaaaaccatctggcctggcctggcctgccccctgagga aggagggggaagctgcagctlgygagagcccttggggcctagactctgttaacatcactatccnatg CACCAAATAATAAACTTTGACGAGTC
DRD2u6	DRD2	G	C	-	-	noncoding	GEI1315	GCACGTGGGTTG GGTGT	GGTCTCCAGG ACTGAAGTT	GCACGTGGGTTGCGGTTCccagcgcctgccccggccttggggaccagcctgacacatgcccttccccaggcggtgttcattcatctgttggtgcccctttctcatcacacatatctgaacatacaact gtgactgcaaacatccccgcctgtccctgtcacgcccctcaactgaggtggcttatgtcaacagcgccc gtgaacccaatcatctacacacaccttcaaatltgatttcgcgaaggccttccctgaagatccctcca ctgctgactctgctgctgcccgcacagcctgctccctcctcctccctgccaggccagccagc cntcaaccttgcgaacctgtagcnaggaaggcctgggtggatcggcctcctcttcannnncccg gaggccccgcagtgctgcctggcctcactgctcctcagtcggcgacacctcactctgcagg i/g c cagtgctagtgaggctgggca tggtaaccagccccctggggctggnccccccagctcaggggcag ctcatagagtcccccctccacacctccagtcaccttcccttggcacaagaatgcagcggccttc cttgaccttcccttggggcttagggttgctgagcctgagtcagggcccccagagctgagtttcc tcttgttggggcttggcgtggagcggcgtggggggagatggagaggttcacacccctgcaaggcc cacaaagcaagcaagctctcttcccaggaacccagAAC TTCAC TCTGGGAGACC

FIG. 5BB

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
DRD2u7	DRD2	C	G	-	-	noncoding	GE1315	GCACGTGGGTGT GGGTGT	GGTCTCCAGG ACTGAAGTT	GCACGTGGGTGTCTCCAGCGTGCCTCCCCGGCTCTGGGGACCAAGCCCTGACCATGCCCTC TCCCCAGGCGTGTTCATCTGCTGGTGGCTCTTCTCATCACACATCTTCAACATACACT GTGACTGCAACATCCCGCTGTCTGTAAAGCGCTCAAGTGGCTGGGTATGTCAACAGCGCC GTGAACCCATCATCAACACCTTCAACATAGTTCCGAAGCCTTCTGAAGATCCTCCA CTGCTACTCTGCTGCTCCCGACAGAGCTGCTTCCACTCCTGCCAGGCCAGCCAGC ONTCAACCTTGCAGAACCGTGAGCAGGAGGCTGGTGGATCGGCTCTCTTCANNCCCCGG CAGGCCCTGCAGTGTCTGCTGGCTCCATGCTCACTGCCCGCACACCTCACTCTGCCAGGG CAGTGTAGTGAAGTGGGATGTACCAAGCCTGGGCTGGGCTGGGCTCAAGTCAAGTCA TAGAGTCCCCCTCCCACTCCAGTCCCCCTATCTTGGCACAAAGATGCAGCGCTCTCTTG ACCTTCTCTGGGCTCTAGGGTGTCTGGAGCCTGAGGCCCCAGAGCTGAGTTTCTTC/
DRD2u8	DRD2	G	A	L	L	cds	GE1088	GAGTGAGGGGT CCCTGG	CTTGGAGGGAG CAGGGG	GAGTGAGGGGTCCCTGGGCTGCACCCCAAGATTCAGGGTCCCCCGCTGGCAGGTACACAGCTG TGGCCATGCCCATGCTG/ATACAATACGGCTACAGTCCAAAGCGCGGTACCGTCTCATGAT CTCCATCGTCTGGTCTCTCTCAACATCTCTGCCACTCTCTCGGACTCAATAAAGCAG GTACATCTCTGCTTTGTGTGGCTGAGGTCAAGTGGCTGGCCCCCTGGCTCCCTCGAAG
DRD2u9	DRD2	A	G	-	-	noncoding	GE1315	GCACGTGGGTGT GGGTGT	GGTCTCCAGG ACTGAAGTT	GCACGTGGGTGTCTCCAGCGTGCCTCCCCGGCTCTGGGGACCAAGCCCTGACCATGCCCTC TCCCCAGGCGTGTTCATCTGCTGGTGGCTCTTCTCATCACACATCTTGAACATACACT GTGACTGCAACATCCCGCTGTCTGTAAAGCGCTTCAAGTGGCTGGGTATGTCAACAGCGCC GTGAACCCATCATCAACACCTTCAACATAGTTCCGAAGCCTTCTGAAGATCCTCCA CTGCTACTCTGCTGCTCCCGACAGCCTGTCTCCACTCCTGCCAGGCCAGCCAGC ONTCAACCTTGCAGAACCGTGAGCAGGAGGCTGGTGGATCGGCTCTCTTCANNCCCCGG CAGGCCCTGCAGTGTCTGCTGGCTCCATGCTCACTGCCCGCACACCTCACTCTGCCAGGG CAGTGTATG/GTGAGCTGGGCTGTTACCAAGCCTGGGCTGGGCTGGGCTCAAGGCGAG CTCATAGAGTCCCCCTCCCACTCCAGTCCCCCTATCTTGGCACCAAGATGCAGCGCTTC CTTGACCTCTCTGGGCTCAGGGTGTCTGGAGCCTGAGTCAAGGCCAGAGGTGAGTTTC TCTTGTGGGCTTGGCTGGAGCAGGCGGTGGGGAGAGATGAGACAGTTCACACCTGCAAGGCC CACAGGAGGCAAGCAGCTCTCTGGCAGGAGCCAGGCACTTCAGTCTCTGGGAGACC
DRD3u1	DRD3	A	G	S	G	cds	GE1135	CAAGCCCCAAA GAGTCTGAT	CGTCAACATGC ACCTGA	CAAGCCCCAAAGAGTCTGATTTATTAATATGTTTCTCTCTCAAGGAGGCCCTTGGCA TCAAGCACTCTCTGGGTATGGCATCTCTGAGTCAGCTGATGATG/AGTCCACTGAACAC CTGTGGGGAGAGAACTCCAGGTGCCAGCGCCCGCCCACTGCTACTACTGCTCTCTCT ACTGCGCTCATCTCGGCATCTGTCTGGCAATGGCTGTGTGTCATGGTGTGTGTAAGGAG CGGCCCTGCAGACTACCACTACTTGTAGTGTAGCCTGGCTGTGGCAGACTGTGTGGTGGC CACCTTGTGTATGCCCCGTGGTATACCTGGAGGTGAGTAGACTTCAGGTGCTATGTGACG

FIG. 50C

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
DRD5d2	DRD5	C	T	H	H	cds	GE1174	GCCATCAAGAT CGTGACCTA	CTGTGACGGT CTGTGCGGT	GCATCATGATCGTGACCTACACGCGCATctaccgcatctaccgcatcgcccgaggcgccagatcgccgaggatcttc ctccctggagagggcgccagagcagcgcagagctgcggagagcagcagcagcctgcggcgcgcgcgaca ccagcctgcgcgcttccatcaagaagagaccaaaggtctcaagacacctgtcggtgatcatgggg gtctcggtgtgttgcggctccctctctcatctcttaactgcatggtcccttctcgagtgaca ccccaggcccttcggcggcttccctgcgtcagtgagaccacctcgacctctcgctctggt tcggctgggctaaactctcaactcaaccccgctcatctatgccttcaacgacgacttccagaagggtg tttgcacagctgctggggtgcagccacttctctccgcacgcccgtggagagcggtgacacatcag caatgagctcatctctcaacaacagacatcgcttctcaaaaggaaatcgacgctgctacatcc a[c/t]atgatgccccacgcggttacccccggcaacccggagggtggacaacgacgagagggagg tctttcgatgcgcatggttccagatctcatcagagctcccgacatggtgacctgtgtcgagctcg tctggagctggactgcgaggggagatcttctttagacaaaataaacaccttccacccgaaatgga ttccattaaactgcattagaaacccctcatggatctgcataACCGCACAGACACTGACCAAG	778
DRD5u1	DRD5	C	T	N	N	cds	GE1171	CCAGGCAGCAA CGGCAC	CTGGGCGATGC GGTAGAT	CCAGGCAGCAAACGGCACGcgctaccgctaccgctctataccagcagctggggcaggggaa cgccgtggggggctcgccgggggacgcgcacactggggccctcacaggtggtcacccgctgcctgc tgacctactcatctctggacctgtctgggcaa[c/t]gtgctggtgtgctgcgagccatcgtgcg gagccgcaacctgcggcccaacatgaccaacgtctcatcgtgtctctgcccgtgtcagaacctt tcgtggcgtgctggttcacgtccctggaggcagtcgcgcaggtggccgggttactggcccttctgga gcgttctgcgacgtctgggtggccttcgacatcatatgtgtccacctgcctccatccctgaacctgtg cgtcatcagcgtggaccgctactggccatctccagcccttccgctacaagcgaagatgactc agcgaatggcccttggatgtgtgctggcctggacatggacctgtccatctcatctcatctccg gtccagctcaactggcacaggacccagcgccctcttggggcgggctgggacctgccaacaacct ggccaaactggagccctggagggaggttctggggagcccgacgtgcatcagagaaactgtgact ccagcctgaatcgaaacctacgcatctcttctctgctcatcagcttctacatccccgttggccatc atgactgactacacgcgcatCTACCGCATCGCCACG	754
DRD5u10	DRD5	G	A	P	P	cds	GE1171	CCAGGCAGCAA CGGCAC	CTGGGCGATGC GGTAGAT	CCAGGCAGCAAACGGCACGcgctaccgctaccgctctataccagcagctggggcagggcaggg ggaaagccgtgggggctcgccgggggacccgcacactggggccctcacaggtggtcacccgctgc ctgctgacctactcatctctggacctgtctgggcaacgtgtggtgtgtgctgcgagccatcgtgcg gagccgcaacctgcggcccaacatgaccaacgtctcatcgtgtctctgcccgtgtcagaacctt tcgtggcgtgctggttcacgtccctggaggcagtcgcgcaggtggccgggttactggcccttctgga gcgttctgcgacgtctgggtggccttcgacatcatatgtgtccacctgcctccatccctgaacctgtg cgtcatcagcgtggaccgctactgggcatctccagcccttccgctacaagcgaagatgactc agcgaatggcccttggatgtgtgctggcctggacatggacctgtccatctcatctcatctccg gtccaaactggagccctggagggaggttctggggagcccgacgtgcatcagagaaactgtgact ccagcctgaatcgaaacctacgcatctcttctctgctcatcagcttctacatccccgttggccatc atgactgactacacgcgcatCTACCGCATCGCCACG	754

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
DRD5u17	DRD5	T	G	V	V	cds	GE1171	CCAGGCAGCAA CGGCAC	CTGGGCGATGC GGTAGAT	CCAGGCAGCAACGGCACGcgctacccggggcgagttcgctctataccagcagctggtgctgaggggaa cgccgtggggggtctgggggggggacccgacactggggccctcacaggtggtcacgcctgcccgc tgacctactcatctggacccctgctggcaacgtgctggtgtgcgacgccaatgctgctggagc cgcaacctggcgcccaacatgacaaacgtcttcactggtgtggtgctggtgctgagacacttttctg ggcgctgctgtctatgcccctggagcgagtcgacgaggtggccggttactggcccttggagcgt tctggacgtctgggtggccttcgacatcatgtgctccacgctccactccactgaaacctgtggctc atcagcgtggacgctactggggccatctccaggcccttcggtacaaagcgaagatgactcagcg catggccttggctatggctggcctggacatggaccttgcactctcatctccctcatctccgttcc agctcaactggcacaggacccaggggccctcttggggcggggtggaacctgccaacaacactggcc aactggacgcccctggagggaggaacttttggagcccgacgtgaatgagagaaactgtgactccag cctgaatcgaaactacgacatctctctctgctcatcagattctacatccccgttt/g/gccatc atgatcgtgacctacacgctgctacccggggcgagttcgctctataccagcagctggcgaggggaa	754
DRD5u18	DRD5	T	C	L	L	cds	GE1171	CCAGGCAGCAA CGGCAC	CTGGGCGATGC GGTAGAT	CCAGGCAGCAACGGCACGcgctacccggggcgagttcgctctataccagcagctggcgaggggaa cgccgtggggggtctgggggggggacccgacactggggccctcacaggtggtcacgcctgcccgc tgacctactcatctggacccctgctggcaacgtgctggtgtgcgacgccaatgctgctggagc cgcaacctggcgcccaacatgacaaacgtcttcactggtgtggtgctggtgctgagacacttt/g/c/t tcgtggcgctgctggtctatgcccctggaaaggcagtcgcccgggtggccggttactggccctttggga ggctctgagcgtctgggtggccttcgacatcatgtgctccactgctccactccactgaaacctgtg cgtcatcagcgtggacgctactggggccatctcacaggcccttcggtacaaagcgcaagatgactc agcgcatggccttggctggtgctggcctggcctggacatggaccttgcactctcatctctcatccg gtccagctcaactggcaacgggacacggcgccctcttggggcggggtggaacctgccaacaacact ggccaaactggacgccctggagaggagacttttggagcccgacgtgaatgagagaaactgtgact ccagcctgaaatcgaaacctacgccatctctctctgctcatcagcttctacatccccgttggccatc atgatcgtgacctacacgctgctacccggggcgagttcgctctataccagcagctggcgaggggaa	754
DRD5u19	DRD5	G	A	G	E	cds	GE1171	CCAGGCAGCAA CGGCAC	CTGGGCGATGC GGTAGAT	CCAGGCAGCAACGGCACGcgctacccggggcgagttcgctctataccagcagctggcgaggggaa cgccgtggggggtctgggggggggacccgacactggggccctcacaggtggtcacgcctgcccgc tgacctactcatctggacccctgctggcaacgtgctggtgtgcgacgccaatgctgctggagc cgcaacctggcgcccaacatgacaaacgtcttcactggtgtggtgctggtgctgagacacttttctg ggcgctgctggctatgcccctggaaaggagtcgcccgggtggccggttactggccctttg/g/a/a ggcttctgagcgtctgggtggccttcgacatcatgtgctccactgctccactccctgaacctgtg gtcatcagcgtggacccgctactggccatctccagcccttcgctacaaagcgcaagatgactc agcgaatggccttggctatggctggcctggacatggaccttgcactccctcatctctcatctccg gtccagctcaactggcacaggacccaggccgctcttgggggggggtggaacctgccaacaacact ggccaaactggacgccctggagaggagacttttggagcccgagcgtgaatgagagaaactgtgact ccagcctgaaatcgaaacctacgccatctctctctgctcatcagcttctacatccccgttggccatc atgatcgtgacctacacgctgctacccggggcgagttcgctctataccagcagctggcgaggggaa	754

[illegible]

40/178

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
DRD5u4	DRD5	C	T	F	F	cds	GE1171	CCAGGCAGCAA CGGCAC	CTGGGCGATGC GGTAGAT	CCAGGCAGCAACGGCACCGcgtaaccggggcaggttcgctctatataccagcagctggcgaggggaa cgccgtgggggctcggcgggggcaccgcccactggggccctcacaggtggtcaccgctgcctgc tgaccctactcaatcatctggaccctgctggcaacgctgctgggtgtgagcagccatcgtgcggagc cgccacctggcgcccaacatgacccaacgtcttcacatcgctctctgctggcggtgcagaccttttcgt ggcgctgctggtcatccctggaaaggcagtcgcccgggttactggccctttggagcgt tctgcagcgtctgggtggcctt[c/t]gacatcatgtgtcctcactgcctccatcctgaacctgtg cgctcatcagcgtggaccgtactggccatctccaggcccttcgctacaaggcgaagatgactc agcgcatggccttgggtcatggctggccctggacatggacctgtccatcctcctccttccatccg gtccagctcaactggcacagggaccagggcgctcttggggggggctggacctgccaacaacct ggccaaactggacgcccctggagggaggaacttttggagcccgcgctgaatgcagagaactgtgact ccagcctgaatcgaaacctacgccaatctctctcctcgtcctacacagcttctacatccccgttgccatc atgatcgtgacctacacgcgcatCTACCGCATCGCCCG	754
DRD5u5	DRD5	C	A	A	A	cds	GE1171	CCAGGCAGCAA CGGCAC	CTGGGCGATGC GGTAGAT	CCAGGCAGCAACGGCACCGcgtaaccggggcaggttcgctctatataccagcagctggcgaggggaa cgccgtgggggctcggcgggggcaccgcccactggggccctcacaggtggtcaccgctgcctgc tgaccctactcaatcatctggaccctgctggcaacgctgctgggtgtgagcagccatcgtgcggagc cgccacctggcgcccaacatgacccaacgtcttcacatcgctctctgctggcggtgcagaccttttcgt ggcgctgctggtcatccctggaaaggcagtcgcccgggttactggccctttggagcgt tctgcagcgtctgggtggccttgcagacatcatgtgtcctcactgc[c/a]tccatcctgaacctgtg cgctcatcagcgtggaccgtactggccatctccaggcccttcgctacaaggcgaagatgactc agcgcatggccttgggtcatggctggccctggacatggacctgtccatcctcctccttccatccg gtccagctcaactggcacagggaccagggcgctcttggggggggctggacctgccaacaacct ggccaaactggacgcccctggagggaggaacttttggagcccgcgctgaatgcagagaactgtgact ccagcctgaatcgaaacctacgccaatctctctcctcgtcctacacagcttctacatccccgttgccatc atgatcgtgacctacacgcgcatCTACCGCATCGCCCG	754
DRD5u6	DRD5	C	G	V	V	cds	GE1171	CCAGGCAGCAA CGGCAC	CTGGGCGATGC GGTAGAT	CCAGGCAGCAACGGCACCGcgtaaccggggcaggttcgctctatataccagcagctggcgaggggaa cgccgtgggggctcggcgggggcaccgcccactggggccctcacaggtggtcaccgctgcctgc tgaccctactcaatcatctggaccctgctggcaacgctgctgggtgtgagcagccatcgtgcggagc cgccacctggcgcccaacatgacccaacgtcttcacatcgctctctgctggcggtgcagaccttttcgt ggcgctgctggtcatccctggaaaggcagtcgcccgggttactggccctttggagcgt tctgcagcgtctgggtggccttcgacatcatgtgtcctcactgcctccatcctgaacctgtgctt c/g]atcagcgtggaccgctactggccatctccaggcccttcgctacaaggcgaagatgactc agcgcatggccttgggtcatggctggccctggacatggacctgtccatcctcctccttccatccg gtccagctcaactggcacagggaccagggcgctcttggggggggctggacctgccaacaacct ggccaaactggacgcccctggagggaggaacttttggagcccgcgctgaatgcagagaactgtgact ccagcctgaatcgaaacctacgccaatctctctcctcgtcctacacagcttctacatccccgttgccatc atgatcgtgacctacacgcgcatCTACCGCATCGCCCG	754

FIG. 5II

[illegible]

FIG. 5J

[illegible]

FIG. 5KK

[illegible]

FIG. 511

[illegible]

FIG. 5MM

45/178

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
F11u3	F11	A	G	T	T	cds	GE287	TGACTTTACTTT TCTCTAGGNGC TG	ACCTCTCCTCA GCATGTATC	TGACTTTACTTTCTCTAGGCTGTGTAAGAAATGTTTCTGATGATATATTTCTACTT ccctttgtttttttagaatactttgtctcttaaacatctgagagtggtatgcccagtacfa /g)cgcatataaaagagcaagctcttctgtttcagttctacaaagctgagagcagatccc aggtaaactgagagttctgcatctctggtgagagtgaccagccccgagaggtctGATACATGCTG AGGGAGGGT
F11u4	F11	A	T	H	L	cds	GE916	TGGAAGGAAG ATGTAGGAAGC	GCAATAAGAC AATCTAATTGG TTAAAGTA	TGGAAGGAAGATGTAGGAAGCTGCTCATCAATGCTTGTGTCAGAGTGTaccaccaaaatc aagccagagatcgcttggagaaactgctgtgtgagtgagtgccgagtgagtgacccctgca cacaactcaccctcagagacacctgctgagaggtccatcttggaacacagtgagatataa cagccgctc[a/t]ctgtttctatgggtcagtagcaccagcgtcttttattagttcatcttctca cacattataaaaaatattactagcatgttaggaaataaaaTACTTTAACCAATTAGATTGCTCTTA TTTG
F11u5	F11	A	T	I	F	cds	GE339	GCCACACACTT CACAAATGTC	CAGGCCGTAAG TCTAGTAGTGT TAAA	GCCACACACTTCACAAATGTCtggaattatttttagtaaaaggaattttcttccctctgtgtt gctcttagggtagagtcacctaaagattttgctgtgtctacagtgagtgcatcttaaatcaatctgaaa taaaagggacacatcttcttcttgggttcaagaaataataatccatgagtaataaaatggca gaaaggggtatgat[a/t]ttgctttgttgaactggaaacacacagtgaaattacacaggtacgg agaaatttataccggaagtgtctccaatggtgaactggataaaaatgTTAACCACTACTAGACTT ACGGCCTG
F11u6	F11	A	T	E	V	cds	GE324	CCTTTATGAGA TTACCACTTAA CTAGA	TTTAAATAATCT GTCTCCTCGAT GT	CCTTTATGAGATTACCACTTACTACAAACaaacaaatcttctcagacaaataacacataact actacatcacag[a/t]atgtgtgactcagttgttgaaggacacctgcttgaaggagggacat tactacggtcttcaacaaagcgaagtagtccaggtagctgactgactacacccaagatgtt tactctcaacttcaacggcgaatcaccatctgagyaatccacccgaaggtaaatgttctgtt ctACATCGAGGAGACAGATTTTTAA
F11u7	F11	C	T	D	D	cds	GE283	GTATTGTGTAT GGTTATCTTAC AAACG	AAAACCCCA CGCATTAAG	GTATTGTGTATGGTTATCTACAAACaaacaaatcttctcagacaaataacacataact ctccagaagccaaagatacccttagtaccacaaagagtgccagaaagatacagaggaacataa aataaccataaagatgactgtgctgggtacaggaagaggaagga[c/t]gcttgcaaggta acagagttctttagccaatggaatataatgcaaatggaaatgCTTAATGGCTTGGGCTTTT
F11u8	F11	G	A	E	E	cds	GE352	TGAGCAAGATG TGCTGAGAT	AGCATGCTGGC ACAGTGAA	TGAGCAAGATGTGCTGAGATggaaggtctgagttgactgtgcaaccttcttgtctccct cgttctagggagattcgggagggcctctgctcctgcaacacaaatgaggtctggcatctggtagggc atcacagagctggggcgaaggtgtgtcacaaggagcggcgaaggtttacacccaacgtgtcga [g/a]tactgtgactggtcttggagaaactcaagcagtgtaaatgggttcccaggggccaattg gagtcctgaaggacccaggaatttctgggagaggggttgagTTCACCTGTCACGATGCT CCTTTATGAGATTACCACTTACTAGATgtagtccagtaaaatccaaataacgcagtcacatgt actacatcacagaatggtgactcagttgttgaaggacacctgcttgaaggaggggacattact acggtcttcaacacaaagcgaagtagtccaggtgtagtctgcaacttaccaccaagatgttcacl t/c]cttcaacttcaacggcgaatcaccatctgaggatccccacccgaatggtaaatgcttctgtt ctACATCGAGGAGACAGATTTTTAA
F13A1d 24	F13A1	C	T	-	-	noncoding	GE284	TGCCATGAATC TTGCAGTATC	CAACTTTTAGC TTACTCTTTCA TGTC	TGCCATGAATCTTGCAATCTggaagaattggagatgacaaactcaca[c/t]tgcccttccct ctgtgaaatgcaggaatgtagtctggtggcccccctcctggttcaagccatcaagcagggccatgt ctgttccaaatttgatgcaacttttcttcttctgagaggtgaagcaggaagctggaggaagtgctg ttttctcccccaacctttaaCACATGAAGAGTAAAGTAAAGTTG
F13A1d 25	F13A1	T	C	-	-	noncoding	GE284	TGCCATGAATC TTGCAGTATC	CAACTTTTAGC TTACTCTTTCA TGTC	TGCCATGAATCTTGCAATCTggaagaattggagatgacaaactcaca[c/t]gccccttccct ctgtgcaatgcaggaatgtagtctggtggcccccctcctggttcaagccatcaagcagggccatgt ctgttccaaatttgatgcaacttttcttcttctgagaggtgaagcaggaagctggaggaagtgctg ttttctcccccaacctttaaCACATGAAGAGTAAAGTAAAGTTG

FIG. 5NN

[illegible]

FIG. 500

[illegible]

FIG. 5PP

[illegible]

FIG. 5QQ

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
F13A1u 8	F13A1	T	C	D	D	cds	GE344	GCCCTACAAAG AAGGGTTTTT	AGAACAGAAAC ATCAGATTGAG TCTA	GCCCTACAAAGAGGGTTTTTcactctgtgtttattaaatgctggtgatgtgtttagctgtggtctgtctctctgtgtgttttacgatgcttggaaatcccgcaagaattgttaccatatttctctgcccattg t/c aatgatgccaatttgcaaatggacattctctctggagaagatgggaaactgtgaaatcccaactcaccagattcagtggtgagtttgattggaattggcaattggctgagtcacattatagtaagtgttacacatttaccacagaagTAGACTCAATCTGATGTTTTCTTCT
F13A1u 9	F13A1	C	A	P	T	cds	GE487	ACACGGTGCAT CCATTA	CGAGTCTCACA AAGAACCA	ACACGGTGCATCCATTAatgtgactttctctctgtggtgattgtatttttgctgtcattatctctggatctccccagggtcaagaagaagagagattggccctagaaactgccctgattgtacggagctaaagaacccctcaacacagaagggtgtcatgaatcaagggtcgaacttgacatttgaggtggaatgtgctgtggtggaaaagacttcaagctctccatcactctccggaacacagccacaacggttacaccatcacagcttattctctcagccaacatcacctctcacccgggttc c/a cgaaggcagaaattcaagaaggagacgttcgacgtgacgtggagccctgtctctgtaagctaacaaatgggtgTTCCTTTGTGAGACTCG
F13Bd2 0	F13B	T	G	-	-	noncoding	GE264	AAAATTGAAAG GCTGAGATTGT AA	CAANTATTAA GCAAGGAAAAA CTCC	AAAATTGAAAGGCTGAGATTGTAA t/g taacacctgactgcaattgatgcttatttcaaaatctctctctctccctcaagaactctgtcttatacaagaaccccttaagaacatagaaatgaatggcagaagaggagctcatatttcaatcatcatgaatctcttataaaatatataatttggaggaaladgtaaacttcGGAGTTTTTCTCTTGGTTAAATATTG
F13Bd2 1	F13B	A	T	E	V	cds	GE370	CTTGAAGAAGC TTTGCTAAAT G	GCCTAAGCAGT GGTCTTTTCTCT A	CTTGAAGAAGCTTTGCTAAATGaaatctgcatgtgtagctaaatggctcagttctcttccaactgtaccttttcagaaggacaggaaggtagcctgtgaggaaaccccttcattgtgaaatgggtgcagcaaatctacactcaagatttattacattggaatggaacacatctgcatgtgaaagcggtctacctctccatggatcgaaatgagataacttgaatcgtggaaaatggacactctctctctgla/t gtgtgtgtgtgtgtgtacatttaccatcagtagctaaacttatggtgtaaaatttctccattctgttactttaattctgaataattctctTAGGAAAGACCCTGCTTGGC
F13Bd2 2	F13B	A	G	Q	Q	cds	GE373	CCAAAATGAAA TCGCCAATAAT A	TTCTTGCAATTG TAGACATAATG A	CCAAAATGAAATGCCAAATAATAcattatttggctcaattttacaatttagtaaaagacagcttagttcatcattaaagttaaataattttttcccatagaaaaatgcactaagcctgagctagtaagtgttacattctgattgtaaaagttattgtataaaatcaagagaacatgcatattggttgcgcttcagggtacaaaacccactggagggtgaaagattgaaagtgttcaatgtctctctgaaggatggctctcacaacccctgtaggaaagaacatggtaagaatcatcttctaaancgtgaaataagtgcttatcaagcttatattgaaatatattgaagcttatattgatactattatttaattagaaattttTCATTATGTTTACAAATGCAGGAA
F13Bd2 3	F13B	T	C	-	-	noncoding	GE373	CCAAAATGAAA TCGCCAATAAT A	TTCTTGCAATTG TAGACATAATG A	CCAAAATGAAATGCCAAATAATAcattatttggctcaattttacaatttagtaaaagacagcttagttcatcattaaagttaaataattttttcccatagaaaaatgcactaagcctgagctagtaagtgttacattctgattgtaaaagttattgtataaaatcaagagaacatgcatattggttgcgcttcagggtacaaaacccactggagggtgaaagattgaaagtgttcaatgtctctctgaaggatggctctcacaacccctgtaggaaagaacatggtaagaatcatcttctaaancgtgaaataagtgcttatcaagcttatattgaaatatattgaagcttatattgatactattatttaattagaaattttTCATTATGTTTACAAATGCAGGAA
F13Bd2 4	F13B	T	A	-	-	noncoding	GE373	CCAAAATGAAA TCGCCAATAAT A	TTCTTGCAATTG TAGACATAATG A	CCAAAATGAAATGCCAAATAATAcattatttggctcaattttacaatttagtaaaagacagcttagttcatcattaaagttaaataattttttcccatagaaaaatgcactaagcctgagctagtaagtgttacattctgattgtaaaagttattgtataaaatcaagagaacatgcatattggttgcgcttcagggtacaaaacccactggagggtgaaagattgaaagtgttcaatgtctctctgaaggatggctctcacaacccctgtaggaaagaacatggtaagaatcatcttctaaancgtgaaataagtgcttatcaagcttatattgaaatatattgaagcttatattgatactattatttaattagaaattttTCATTATGTTTACAAATGCAGGAA

FIG. 5RR

[illegible]

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
F13Bul	F13B	A	T	-	-	noncoding	GE358	GATGCTTGACA COATGAATATT TTA	GTACAGTTGAA AGAGAAGTAC CA	GATGCTTGACACGATGAATATTTAatttaatttattatttttttggtaattatatgactaaagg tgctgatta[a/t]tttttccataaggaagaacacagatgtcctcctccacctctgcccata aactccaaaattcaaacacattcaaacattatcgtcatgggaaatagttcatatagaaatgga acttaattttgagatccatgggtcagcagaataatcgttggagagatggaaaaatcgacagaacctc caaaaatgcatgggttagtaaaccttgaagaagccttggctaaaaatgaaatcctgcatgtgtagcta aatGGTCAGTTCTCTTCAACTGTAC	351
F13Bul 0	F13B	C	T	N	N	cds	GE365	CAACACAACGA TTCTGTTCTTT	GAGTGGTATAG AACATAACATT TCTGA	CAACACAACGATTCCTGTTCTTaccagaatgaacaagatgtttcttcttttcttcttctt tttttaattgtgtgtccagagccatgcacattatcttttactgaaatggaaaaaataattttac ttctgaaatgggatttttgacaa[c/t]agaccacacatttttgatgggtgaatatattgagtttat ttgtagaggagatacttctccagctgaattatataattactggatctatacttagaatgcaatgtg acagagggcagttaaaatatccaaagatgtattccaaagacaaagggttaagaagtttttttttgg tcagattatttttttgggtcagattgttattcaacattTCAGAAATGTTATGTTCTATACCCTC	390
F13Bul 1	F13B	T	C	G	G	cds	GE396	TGTTGGATGTT TAAAGTCATTT GC	CATTTTATTG GACCCCTATTT T	TGTTGGATGTTTAAAGTCATTTGCGagatcaattatgataaaaggactccttgagttgtcacaaaa gtaccttaaaatttaagtaagaaataaagtaactagttgaagtgctctctaaaaacttttagttt gtattgtcacctgattacaaatttatgttttttagattttgttttccatatttcagcttgacattta ctcatttcagagaaacccctgtggttttccctcatgtggaaatggaaatggcccaatatattacta tacttttaaaagcttttaactttccaatgagcatagacaaaattgtcattttctgtcttggtg g[t/c]tatacacctgaagtggaagacaaagaaagacaaacacgtgtacaacagaaggctggct tcagagagccaaagtgcttcagtaagtcagctggatattgtcacctcaatgttttcaataactcaagaa atttgtatataaaaaATAGGGGTCCCAATAAATG	489
F13Bul 2	F13B	A	G	H	R	cds	GE370	CTTGAAGAAGC TTTGCTAAAT G	GGCTAAGCAGT GGTCTTTTCTCT A	CTTGAAGAAGCTTGTCTGTAGCACTTAAATGaaatctgcatgtgtagctaatggctcagttctcttcaactg taccttttcagaaggacaggggaaggttagctgtgaggaaccccttctcattgaaaaatgggtgcag caaaatttac[a/g]ctctaagattttattacaatgggataaaatgacatatgcattgtaaaagcgg ctaccttccctcagatcgatgagataaactgtgaactgtgaactgtggaatggacacttctcctgagt gtgttggtatgtatgctacatttaccatcagtagctaaacttatgggtgtaaaaaatttccattctg tctcttaactcgaatatcttcTAGAAAGACCACTGCTTAGGC	369
F13Bul 3	F13B	A	T	-	-	noncoding	GE380	GGAGACTCTGT CTCTGTAGCAC TTAT	AACACAATGT AGCAATATATAG CATT	GGAGACTCTGTCTGTAGCACTTAAagtaactggatgttccattatagcaattcattgtatact ttaaaacttatttttgc[a/t]gaatctaaaggaatgtgcacatctcctcctctcttattaaacatg gagtcattattagttcaacagtagacaccttgaaatggcttccagtagaatacagatgtttt gatcaccaatttccctagaaggatctaggaggcctattgttttagatggaatgtggactacaccacc attgtgttttaggtatgtactactaaatagcctctaaacaaagtaaaactatatttttaatttg ctgtcattttttggagtttaacatacactaatatAATGCTATATTGCTACATTGCTGTT	383
F13Bul 4	F13B	T	G	D	E	cds	GE380	GGAGACTCTGT CTCTGTAGCAC TTAT	AACACAATGT AGCAATATATAG CATT	GGAGACTCTGTCTGTAGCACTTAAagtaactggatgttccattatagcaattcattgtatact ttaaaacttatttttgcagaatctaaaggaatgtgcacatctcctcctctcttattaaacatggagt cattattagttcaacagtagacaccttgaaatggcttccagtagaatacagatgttttgatc accatttccctagaaggatctaggaggcctattgttttaga[t/g]ggaaatgtggactacaccacc attgtgttttaggtatgtactactaaatagcctctaaacaaagtaaaactatatttttaatttg ctgtcattttttggagtttaacatacactaatatAATGCTATATTGCTACATTGCTGTT	383
F13Bul 5	F13B	A	C	Y	S	cds	GE380	GGAGACTCTGT CTCTGTAGCAC TTAT	AACACAATGT AGCAATATATAG CATT	GGAGACTCTGTCTGTAGCACTTAAagtaactggatgttccattatagcaattcattgtatact ttaaaacttatttttgcagaatctaaaggaatgtgcacatctcctcctctcttattaaacatggagt cattattagttcaacagtagacacct[a/c]tgaaatggcttccagtagaatacagatgtttt gatcaccaatttccctagaaggatctaggaggcctattgttttagatggaaatgtggactacaccacc attgtgttttaggtatgtactactaaatagcctctaaacaaagtaaaactatatttttaatttg ctgtcattttttggagtttaacatacactaatatAATGCTATATTGCTACATTGCTGTT	383

FIG. 5TT

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Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
F13Bu1 6	F13B	T	C	P	P	cds	GE370	CTTGAAGAAGC TTTGCTAAAAT G	GCCTAAGCAGT GGTCTTTTCT A	CTTGAAGAAGCCTTGTCTAAATGaaatctgcatctgtagctaaatggtgcagttctcttcaactg tactcttcagagagacagagaggttagctgaggaacacaccccttcatgaaaaatggtgcag caaatctacactctaaagtattacaatgggataaaagtacataatgcatgtaaaagcggctac cttctccatggatcgaaatgagataaacttgtaactggaataatggacacttcc{t/c}cctgagt ggttggtatgtagctacattaccatcagtagtaaaacttatggtgtaaaattttccattctg ttactttaactgcaatatctccTAGAAAAGACCTTAGGC	369
F13Bu1 7	F13B	T	A	N	K	cds	GE400	TTTGCTGTCAA CTCTTGCTTAG	AAATGAGAGAG AAACACCTGT GAAA	TTTGCTGTCAACCTTGTCTAGGaaatttttaaaatatattgagctgaaaaaaatttttcatatttg taatgaatttataaaacaattgctctaaattataatacaaaatctcgagacacactatgtagag tactatagtagtattgttatgagctcatattaaattttaaacaacaccccttctcttagaaaaa{t/a }aatgagaatttgaagcatcctcctgttgtaaatgggctgttgcagacgggatatggcaa gctatgcaacagagatcctcagtggaataatagatgcaatgaatatctactgagggatcaaaa atatctcgttgggaacaaggaaaatggtcatccacacccctgttgcctgggtgaagaaagagacac atggaatgtctcagtttctactttttagtatttcttctacagtttcttatcatcatgaaatgatg atttctgtaacaatctcttTTTCACAAGTGTTTTCTCTCTCATTT	498
F13Bu1 8	F13B	G	C	L	L	cds	GE264	AAATTTGAAAG GCTGAGATTGT AA	CAATATTTTAA GCAAGGAAAA CTCC	AAATTTGAAAGGCTGAGATTGTAAataacacccctgactgcaattgtagcttatttcaaaatctct cttttctccctcaagcactct{g/c}tcttcaagaaccccttagaacaacatagaaatgaatggca gaaagagagtagtcatatttcaatacatcatgaaattctctataaaataataatttugaggaaataag tcaaaaactctGGAGTTTTCCTTCTGCTTAAATAATTTG	232
F13Bu1 9	F13B	T	A	N	K	cds	GE400	TTTGCTGTCAA CTCTTGCTTAG	AAATGAGAGAG AAACACCTGT GAAA	TTTGCTGTCAACTCTTGTCTAGGaaattttaaataatttggagctgaaaaaaatttttcatatttg taatgaatttataaaacaattgctctaaattataatacaaaatctcgagacacactatgtagag tactatagtagtattgttatgagctcatattaaattttaaacaacaccccttctcttagaaaaa{ t/a}lgagaatttgaagcatcctcctgttgaatgggctgttgcagacgggatatggcaa gctatgcaacagagatcctcagtggaataatagatgcaatgaatatctactgagggatcaaaa atatctcgttgggaacaaggaaaatggtcatccacccctgttgcctgggtgaagaaagagacac atggaatgtctcagtttctactttttagtatttcttctacagtttcttatcatcatgaaatgatg atttctgtaacaatctcttTTTCACAAGTGTTTTCTCTCTCATTT	498
F13Bu2	F13B	A	G	M	V	cds	GE396	TGTGGATGTT TAAAGTCATT GC	CATTTTATTG GACCCCTATT T	TGTTGATGTTTAAAGTCATTGTCagatcaattatgataaaagagactccttgcagttgtcacaaaa gtaccttaaaaaatttaagtaagaaaaataagtagtactagttgaagtgtctcttaaaaaacttttagttt gtattgtcacctgattacaatttatgttttttagatttgttttccatattcagcttgacattba ctcatctcagagaaacccctggtgttctcctcatgtggaaaaatggaagaatttgcacaaatattacta tacttttaaaagcttttactttcca{a/g}tgagcatagacaaaaaaatttgcatttttctgcttg gctggttataccactgaaagtggagacagaagagcaaacccacggtgtacacagaaggctggctc tccagagcccaagggtgcttcagtaagtgcagctggatgtcactcaatgttccaatactcaaaagaa atttgtatatataaaATAGGGGTCCATAAATAATG	489
F13Bu3	F13B	A	G	H	R	cds	GE373	CCAAATGAA TCGCCAATAAT A	TTCTGTCATTG TAGACATAATG A	CCAAATGAAATGCCAATAATAacattatacttttggcttaatttttacaattttagtaaaagaca agcttagtttctcatcattaaagttaataaattttttcccatagaaaaatgcactaagcctgacct ggtaaatggtttacatctctgtatgaagtatttataaaaaatccaagagaacatgc{a/g}ttat ggttgctgcttcagggtacaaaacacccctggaggagatgaagaagtgttcaatgtctctctga tggatggtctctctcaaccacacccctgtaggaagaagacatggtataagaatcatttccctaaactgaa gataagtgcttatcaagcttataattgaatatattgaagcttatattgatatctattttaaataag aatttattttTCATTATGCTTACAAATGCAGGAA	423

FIG. 5UU

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Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
F13Bu4	F13B	A	T	N	Y	cds	GE358	GATGCTTGACA CGATGAATATT TTA	GTACAGTTGAA AGAGAACTGAC CA	GATGCTTGACACGATGAATATTAAATTAAATCTTATTATCTTTGTAATATGACTAAAGG Tgctgattaaattttcccaaggaagaaacacagatgtctctccacccctctgcccataaact ccaaaatttcaaacacatttcaacaacttctcgtcatgagagaatagttcatalagaaatgtgaactt [a/t]attttgagatccatgggtcagcagaaatcacgttgtgagatggaaaatcgacagaaacctc caaaatgcattgggttagtaacaccttgaagaagcctttgctaaatgaaatcgaatcgtgtgagcta aaTGGTCACTTCTCTTCAACTGAC	351
F13Bu5	F13B	C	T	P	S	cds	GE370	CTTGAAGAAGC TTTGCTAAAT G	GCCTAAGCAGT GGTCTTTTCCT A	CTTGAAGAAGCTTGTGCTAAATGaaatctgcatgtgtgagctaaatggcagttctcttccaaactg taccttttcagaagacagaggaaggttagcctgtgaggaaccccttccattgaaaatgggtgcag caaaatttcaaccttaagatttattacaatggggaataaagtgacataatgcattgaaaagcggctac cttctccatggatcgaaatgagataaacttgaatcgtggaataatggacactt[c/t]ctcctgagt gtgttggtatgtatgctacatttaccatcagtagtcaaaacttatggtgtgtaaaatttccattctg ttacttttaattctgaatatctcctTAGGAAAGAACCACTGCTTAGGC	369
F13Bu6	F13B	T	A	M	K	cds	GE400	TTTGCTGTCAA CTCTTGCTTAG	AAATGAGAGAG AAAACACTTGT GAAA	TTTGCTGTCAACTCTTGTGCTAGCAaaattttaaataattttgagctgaaataattttlccatatttg caatgaattttataaaacaatttgcctaaattataataatacaaatatcgagacactatagtatgag ttactatagatatattgttatgagctcatattttaaacaaccccttcttcttagaaaaataat gagaattgtgaagcatctcctctgttgaatgagtggtgtgtgagacggga[t/a]attggcaa gctatgcaacaggaatcctcagtggaatagatgcaatgaatatttacttactgagggatcaaaa atatctcgttgcgaacaaggaatgggtacccccacctgttgccttgggtggaagaagagaaacac atggaatgctctacgtttgtacttttatgtgattttcttctacagtgttttatcatcatgaaaatgatg attttgtacaaatctttttCACAAAGTGTCTCTCTCATTT	498
F13Bu7	F13B	T	A	I	K	cds	GE400	TTTGCTGTCAA CTCTTGCTTAG	AAATGAGAGAG AAAACACTTGT GAAA	TTTGCTGTCAACTCTTGTGCTAGCAaaattttaaataattttgagctgaaataattttlccatatttg baatgaattttataaaacaatttgcctaaattataataatacaaatatcgagacactatagtatgag ttactatagatatattgttatgagctcatattttaaacaaccccttcttcttagaaaaataat gagaattgtgaagcatcctcctgttgaatgagtggtgtgtgagacggga[t/a]attggcaa gctatgcaacaggaatcctcagtggaatagatgcaatgaatatttacttactgagggatcaaaa atatctcgttgcgaacaaggaatgggtacccccacctgttgccttgggtggaagaagagaaacac atggaatgctctacgtttgtacttttatgtgattttcttctacagtgttttatcatcatgaaaatgatg attttgtacaaatctttttCACAAAGTGTCTCTCTCATTT	498
F13Bu8	F13B	G	A	-	-	noncoding	GE400	TTTGCTGTCAA CTCTTGCTTAG	AAATGAGAGAG AAAACACTTGT GAAA	TTTGCTGTCAACTCTTGTGCTAGCAaaattttaaataattttgagctgaaataattttlccatatttg taatgaattttataaaacaatttgcctaaattataataatacaaatatcgagacactatagtatgag ttactatagatatattgttatgagctcatattttaaacaaccccttcttcttagaaaaataat gagaattgtgaagcatcctcctgttgaatgagtggtgtgtgagacgggaatattggcaagcta tgcacaaggaatcctcagtggaatagatgcaatgaatatttacttactgagggatcaaaaatat ctcgttgcgaacaaggaatgggtacccccacctgttgccttgggtggaagaagaa[t/a]agaacac atggaatgctctacgtttgtacttttatgtgattttcttctacagtgttttatcatcatgaaaatgatg attttgtacaaatctttttCACAAAGTGTCTCTCTCATTT	498
F13Bu9	F13B	T	A	-	-	noncoding	GE380	GGAGACTCTGT CTCTGTAGCAC TTAT	AACACAAATGT AGCAAAATATAG CATT	GGAGACTCTGTCTGTAGCACTTAAaagtaactggatgttcatattatagcaatttcatgtatact ttaaact[t/a]atttttcagaaatctaaaggaatgtgcacatctcctcctctattataaactg gagtcattattagttcaacagtagacacccatgaaaatggctcttcagtagaatacagatgtttt gatcaccatttccctagaaggaatctaggaggccctattgttttagatggaatgtggactacacccc attgtgtttagggtatgtactactaaatattgcctcctcaacaagtaaaactatatttttaatttg ctgtcatttttgagtttaacataacactaatataATGCTATATTGCTACATTTGTGTT	383

FIG. 5VV

[illegible]

FIG. 5YY

[illegible]

FIG. 5ZZ

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')
F5u20	F5	G	A	S	S	cds	GE494	TCTGTCCTCCTT TCTGTAGGAAC T	CCCTGTGACAT CTGGTGTAG
F5u21	F5	G	A	R	H	cds	GE391	CCCCTTCTCAC CAACAAGC	AGCAGGTGAGG CATTCCTGG
F5u22	F5	A	G	R	G	cds	GE391	CCCCTTCTCAC CAACAAGC	AGCAGGTGAGG CATTCCTGG
F5u23	F5	T	A	L	Q	cds	GE316	GCAAAGGTTT AACATCTTCTT T	GCACAGTCTTC AGATTGCTTT
F5u24	F5	A	T	G	G	cds	GE267	TGATGACCCTG AATACAGACAT AGT	AGTTTAAATG TTGATGCTGGT ATT
F5u25	F5	G	T	S	S	cds	GE362	GCTATCCCAGA TTTGAGAGTGG T	TTTGTCCCATG ACAGAACTCC

FIG. 5AAA

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Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
F5u26	F5	C	T	-	-	noncoding	GE924	TTGCTTGGCCCT AAACTCTTTG	GAAACACAGGAC CGAATAATTAC TA	TTGCTTGGCCCTAAACTCTTTGntatccaggaaagntaatgagngntnttttatctgagtgctactgaaacaagtcttcttctcatagggaccctaaactgaggggtgggacacagaagacgttttgacaagcaaatcgtgctactatctgctgtgtttgatgaaagcaagagctggagccagtcacatccctaatgtacacagtcagtgaatgtgaaatgggacaaatgccaggtaaca[c/t]gaggggctgtgtaccatcaacaacagtaaaatcatTAGTAATTTTTCGGTCTCTGTTTC	303
F5u27	F5	A	G	E	E	cds	GE494	TCTGTCTCTCT TCTGTAGGAAC T	CCCTGTGACAT CTGGCTGTAG	TCTGTCTCTCTTCTGTAGGAACCTTggatgttaacttccatgaattcttagtccaagaagcaaaaaagctgagggctgaaattcaggagatgttaaatgtatccagatgatgatgaagactcatatgagatcttttgaaacctccagaaattcagctacagggaaatgcatgatcgttttagaaccttgagagatgaagagtgatgctgactatgattaccagaacagactggctgagcatttaggaattaggtcatttcgaaactcatattgaaccaggaaagaagagttcaatcttactgcccctagctctgggagaatggcaactga[a/g]ttcgtttcttcgaacacagataaattgttggttcaaatattcttccccaaagt aattatagtaagttcaactgtcaataaaccttgcagaacctcagaagaagcccccttctcaccacaagc caccacagctggttccccacitgagacacctcatctggcaagaactcagttctcaattcttccacag cagagcattccagcccatattctgaagaccctatagaggatcctctACAGCCAGATGTACAGGG	585
F5u28	F5	C	T	S	S	cds	GE391	CCCCTCTCAC CAACAAGC	AGCAGTGTAGG CATTTCTGG	CCCCTCTCTCACCAAGCCaccacagctggttccccactgagacacctcatctggcagaactca gttctcaattcttccacagcagagcattccag[c/t]ccatacttgaagaccctatagaggatc ctctacagccagatgtcacaggagatcgtctacttctcacttggctgggagaattcagaagtcaa gaaatgctaagctaaaggacccaaggtagaagaagatacaagcaaaagcacaggttctcctg gatgaattactagcacataaagttgggagacacctaaagccaaagacactggttctccttccggaa tgagggccctgggagaccttctcctagccaagacacactgggttctccttccagaatgagggccctgggag gacctcctagtgatctgttactcttaaaacaaagtaactcatctcaagattttggttgggagatg gcaattggcttctgagaaggttagctatgaataatcccaagatactgatgaagacacagctgtta acaattggctgatcagcccCCAGAAATGCTCTCAGTCT	558
F5u29	F5	A	T	K	*	cds	GE389	GAAAGGTAGCT ATGAATAATC CAAGA	GGGTCTTGAAT GGGGAATGT	GAAAGGTAGCTATGAATAATCCAGATAactgatgaagacacagctgtttaaacaattggctgatca gcccacagaatgctcctcagctgcttggggagaaagacccctcttgcacaacagcctggaaagcag agtggccaccccaagtttctctaggttagacataaatctctacaagtaagacaggtggagggaaa gagtagactgaagaaagccagtttctcattaaagacacgaaaaaaaagaaagagagacacacac accatgctcttcttctcggagaccttccaccttaagaagtgagcctacacacacatttcca gaagaagacttaagcattcgttgggtgcttcat[a/t]aatccaatgaacacatcttctccacag acctcaatcagacattggcctctatggattttgggtggatagcctcacttctgaccataatcag aattcctcaaatgacactggttcaggcaagctgctcctcaggtcttctatcagacagtgccccaga ggaacactatcaaaCATTCCTCCATTCAGACCC	553
F5u3	F5	G	A	R	Q	cds	GE47	GAAATAACTTT GCAATGAAA CA	TTTCTGAAAGG TTACTTCAAGG AC	GAAATAACTTTGCAATGAAAACatttttgaatataatttcttccaggcaggaaacaacccatga tcagagcagttcaaccaggggaaacctataactataagtggaaacattttagagttttagaaccac acagaaaaatgatgccagtgcttaacaagaccatactacagtgacgtggacatcatgagagacat cgcctctgggctaataggactacttctaattctgtaagagcagatccctggacaggc[g/a]agga atacaggtatttcttCTCTTGAAGTAACCTTTTCAGAAA	297

FIG. 5BBB

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
F5u30	F5	C	A	P	T	cds	GE389	GAAAGGTAGCTT ATGAAATAATC CAAGA	GGGTCTTGAAT GGGGAATGT	GAAAGGTAGCTTAAATAATCAAGATactgtgaagacacagctgttaacaattggctgatca gccccagaaatgcctcacgtgcttggggagaagcaccctcttgcacaacagcctggaaagcag agtggccaccacaaagtctctagagtttagacataaactctcaagtaagacaggtggaggaaa gagtagactggaagaaagccagttctcattgaagacacgaaagaaagagagacacacac accatgctctcttattctccgaggaaccttccaccttaagaagtgaagcctacacacattttca gaaagagacttaagcattcgttgggtcttataaatacgaatgaacatctcttccacagacct caatcagacattgcccctctaggttttggctggatagcctcactt(c/a)ctgacataatcag aatcctcaaatgacactggtcaggcaagctgctccaggtcttctatcagacagtgccccccaga ggacacatacaaacattcccatTCAGACCC	553
F5u31	F5	G	A	M	I	cds	GE925	CACTCGCCCTC TCTGTGTC	CAGATTGCCCTT TTCCCTGTATT	CACTCGCCCTCCTCTGTGTGTCaagatttttaattgatttcaactcttngtcnnttcagccattaa tgggatgactctacagcttgcctggcctgaaat(g/a)atgagaagagtggtgggttaccac ctgctgaacataaggcggctcccaagacattcacgtggttcaacttcaaggccagaccttggctgga aaatggcaataaacacagcaccagttaggggtctggcccttctgctggttaagatgggttaattgg gaaggggccctgctaaagaAATACAGGGAAAGGCAATCTG	301
F5u32	F5	T	G	F	V	cds	GE910	GCAGGGAATAT TGGTGTG	TTTACTGATAC AATGCCAGAGT T	GCAGGGAATATTTGGTGTGtaatttaatacacgggaaccacacacatgacaggtgttccacacccctg ggatggaaatggaaagatagaaacaaagcaaatcacagcttctcg(t/g)taagaaatctt ggggggagattactgggaaccttccgtgctgctgaatgccagggagcgtgtgaattgcctgg caagccaaaggccaagtatactatgcatggttctcttaggtctccaaagaaagcaagggcc ctcactaccacaaacatggaggtctgggaagcaggagatttttaactgtttctgcaaatataaa atcctagggtccaanagtcagggtctgctggacttgatanccttgaactataacttctctcaaa aanaagtcatttatactatctctggtgttttgggtctatcttggattaaaccaaataaagcaaat taattttctgcaaggggatanatggttaataattttagtcttctggggccatcacattctctgcca taactactactCTGCCATTTCTATCAGTAA	552
F5u33	F5	A	G	D	G	cds	GE921	AAAAACCTTAG CCATTATGTT GT	AAGAAAGAGAA ATAGTGGAATA C	AAAAACCTTAGCATTTATGTTGTCattaaagatttctcttatttggccttccagatttttgaag gaaatactactaaccaagagcatgtgaagaacttctcaaccccaaatctctccaggtttatc cgtgtcattctataaaacatggaatcaaatggaatttgcacttgcctgggaactcttggctgtg(a/g tatttactagaattgaacattcaaaacccctgggaagagactctttaaagacctcaaacattta gaatgggcaatgtattttaaagctgtgttaaatgttaacagTTTCCACTATTTCTCTTCTT	322
F5u34	F5	A	G	Q	Q	cds	GE912	TCTACTAGAAG TCTGAAGATAT GAGAG	TGCCAAATTTAC CCATAGAAAT	TCTACTAGAAGTCTGAAGATATGAGAGatctacttttttggtaattgagaaatagatatgaat atgctcttcaaatagtttttggatttttatttccagtttgaattcttctgtaacttctttaaaga aaattgtctacagagagtgaaccataattttaaagaaagaaacccaca(a/g)ctaccatttc aggtaagcctgaaatataactttttaaattttaaagataaaATTTCTATGTTTGGTAATTTGGCA	255
F5u35	F5	G	A	A	A	cds	GE362	GCTATCCCAGA TTTGAGAGTGG T	TTTGTCCCATG ACAGAACTCC	GCTATCCCAGATTGAGAGTGTGtaaaacgcaatcctcagaaactgcccacatgtcttgatggct gttactcctccaggtgcttctaccttgaccacacattccctgc(g/a)gagaagatggacgacg ctgtggctccagggccgagaaatcacacctatgaaatgagatcagtgaggacagtggaacccacccat gatgacctccatgctccacacacatctattactcccatgaaatctgacgaggttttcaactc ggggctgattggggccctgcttactctgtaaaaggttaagaaaccccccccccaaaagattcaaca actaaatgttgggaatggtcagGAGTTCTGTCTATGGGACAAA	366
F5u36	F5	G	A	V	M	cds	GE915	TGATTATCAGA AGAGCAAGGAA A	ACTGTGACCCA GTGTGATTTA	TGATTATCAGAAAGCAAGGAAAtccttgagaagagggcaatcacaaatttactctgttttccagg aaaaagatactcactcaggttgataggtccctcctaacttgcacaaaggaata-tacataag gacagcaactgctt(g/a)tggaactgagagaatttcttactactatttactcttggatgaa agaagctggttactatgaaagaaagtcctccgaagttcttggagactcacatctcagaaatgaaa aaatcccatgagtttcaagggtatttttctcctggactttgactcctaatctcctaatTAAATTCACACTG GGTCACAGT	334

FIG. 50CC

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
F5u37	F5	C	T	P	S	cds	GE391	CCCCTTCTCAC CAACAAGC	AGCAGGTGAGG CATTTCTGG	CCCCTTCTCACCAAGCCaccacagctgggtcc(c/t)cactgagacacccctcatgggcaagaa ctcagttctcaattctccacagcagacattccagcccatattctgaagccctatagaggatc ctctacagccagatgtcacaggatacgtctacttctcaacttggctgagagaattcagaagtcaa gaacatgctaagcgttaaggcccaaggtagaagagatcaagcagcaaacacaggttctcctg gatgaattactagcacataaagttgggagacaccccaagcagacactgggttctcctccggaa tgaggccctgggagacacttcttagccaagacacactgggttctcctcagaaatgagccctgggag gacccctcctagtgatctgttactcttaaaacaagaaactcctccttaagatttgggtgggagatg gcaatttggcttctgagaaaggttagctatgaataatccaagatactgatgaagacacagctgtta acaatttggctgatacagcccccAGAAATGCTCAGTCT	558
F5u38	F5	G	C	E	D	cds	GE362	GCTATCCAGATTTGAGAGTGTGaaacgcgaatcctcagaaactgcccacatgtcttgatggct TTTGAGAGTGG T	TTTGTCCCATG ACAGACTCC	GCTATCCAGATTTGAGAGTGTGaaacgcgaatcctcagaaactgcccacatgtcttgatggct gttactcctccagtgcttcttacttgaacacacatttccctggga(g/c)aaatggagcagc ctgtggctccagccgagaaacacacattgaatgagatcagtgaggagacagtggacccaccat gatgaacctccatccctcacacatttactcccatgaaatctgatcagagatttcaactc ggggctgattgggcccctgcttattctgtaaaaaaggttaagacaccccccaaaaagatttcaaca actaaabgltggatgggtcagagTTCTGTCTCATGGACAAA	366
F5u39	F5	G	A	E	K	cds	GE391	CCCCTTCTCAC CAACAAGC	AGCAGGTGAGG CATTTCTGG	CCCCTTCTCACCAAGCCaccacagctgggttcccccactgagacacacttggcagaagaaactca gttctcaattctccacagcagagcattccagcccatattctgaagacccctatagaggatcctct acagccagatgtccagggatacgtctacttctcacttggctggagaattcagaagcaagaac atgctaagcgttaaggcccaaggtagaagagatcaagcagcaaacacaggttctcctggatg aaattactagcacataaagttgggagacacctaagcagacactgggttctcctccgaaatgag gccccgggagacacttcttagccaagacactgggttctcctccagaaatgggcccctgg(g/a)ag gacccctcctagtgatctgttactcttaaaacaagaaactcctccttaagatttgggtgggagatg gcaatttggcttctgagaaaggttagctatgaataatccaagatactgatgaagacacagctgtta acaatttggctgatacagcccccAGAAATGCTCAGTCT	558
F5u4	F5	T	C	M	T	cds	GE172	TTTAAGAAAT ACAGGTCTCAG CAT	TTTCTCCCATG ATTCTGTATT GT	TTTAAGAAATACAGGTCTCAGATTTtgagataatttctcaaacacaaattggaaaaacattataaga aagttatgtacacacagtcagagatgggtccttcccaaacatacagtgatcccaata(t/c) gaaagaagatgggatttgggtcctattatcagagcccggtcagagacacactcaaaagtaagta ACAAATACAGAAATCATGGGAGAA	221
F5u40	F5	C	G	P	A	cds	GE391	CCCCTTCTCAC CAACAAGC	AGCAGGTGAGG CATTTCTGG	CCCCTTCTCACCAAGCCaccacagctgggttcccccactgagacacacttggcagaagaaactca gttctcaattctccacagcagagcattccagcccatattctgaagacccctatagaggatcctct acagccagatgtccagggatacgtctacttctcacttggctggagaattcagaagtcagaac atgctaagcgttaaggga(c/g)ccaagtagaagagatcaagcagcaaacacaggttctcctg gatgaattactagcacataaagttgggagacaccccaagcagacactgggttctcctccggaa tgaggccctgggagacacttcttagccaagacactgggttctcctccagaaatgagccctgggag gacccctcctagtgatctgttactcttaaaacaagaaactcctccttaagatttgggtgggagatg gcaatttggcttctgagaaaggttagctatgaataatccaagatactgatgaagacacagctgtta acaatttggctgatacagcccccAGAAATGCTCAGTCT	558
F5u41	F5	C	G	L	V	cds	GE915	TGATTATCAGA AGAGCAAGGAA A	ACTGTGACCCA GTGTGATTTA	TGATTATCAGAAGAGCAAGGAAAttcctgagaaaggggcaatacaatttactctgttttccagg aaaaagatatctcactcaggttgataggccctc(c/g)taactctgcaaaaaaggaata-taca taaggacagcaacatgccttagacatgagagaaatttcttactattttagacctttagtgaaa agaagagctgggtactatgaaaagagtcgggaagtctcttgagagactcacatcctcagaaatgaaa aaatcccatgaggttccaggtatttctcctggacttctgactctcctcctaatTAATACACTG GGTCACAGT	334

FIG. 5DDD

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
F5u42	F5	T	G	Y	D	cds	GE279	GAATTTAGGCA GTGTGTGACIT G	AGAGATTCAGA TAGAATATGCG ACAC	GAATTTAGGCAGTGTGTGACTGTGtgacaaggacaggttctgtttactggcttctctataattgcag gtggacatgcaaaagggaagtcatataatcacagggtccagagcccaagggtgcacaaactaactactgaa gtcctgctataccacagagttct/gt/gatgagcttaccagttccaaaccagatcaactggagatc ttcaaaagggaacagcacaaaggaaatgtagtggtttGTGTGCATATTTTCTATCTGAATCTCT
F5u44	F5	C	A	L	I	cds	GE395	GACCTCAGGCA GACAAACCT	TCTGATCAAGG TCTGGAGGA	GACCTCAGCCACAAACCTctctccagaactcagtcagacaacaccttctctagaccttcagccagacaacacctct gatgcccccttctccagacatcagcccatcaaaccttctctagaccttcagccagacaacacctctct ctccagaactcagccatagctctctccagaactcagtcagacaacaccttctccagccccctc ggtcagatgccccatttctccagacctcagccatacaacc[c/a]tctctctagaccttcagccaga caacctctctccagaactcagtcacaacaccttctccagccccctcagtcagatgcccccttct ccagacccccagcatcaaaccttctctagacctcagccagccagacaacacctctctccagaactcag tcagacaacaccttctccagacctcagtcagatgcccccttctccagaccttcagtcagtcacaactcccc ttacccccagacctcagacagatgacaccttctccagaccttggtagagacagatcttctcccaaac tttggtcagatgcccccttcccaagacctcagccagtgacctctctctccagacatcagtgacac caccttctccggatctcagccagatatacctTCTCCAGACCTTGATCAGA
F5u45	F5	T	A	L	I	cds	GE923	GATCATTCCTT TTCCTAGGTT	TTCAGATTACG AGGTTAGGGGA	GATCATTCCTTTTCTAGGTCgttttbaaaat[t/a]tagcatccagacctatctctcacatg cccatggaccttctctatgaaaaatcatcagagggaagacctttagagatgacctctctctacatgg tttaagggaagtataatgctgttcagccaaatagcagttatacctacgtagtgatgacctgccactgagcg atcagggccagaaagtctcgctctgctgctgggcttgggacctactactcagctgtgaaacccag tagtactcttctcatgaaagttttctctcatTCCCTAACCTCGTAATCTGAA
F5u46	F5	C	A	L	I	cds	GE387	GTGCCCCCAGA GGAACACTA	TCATATGGCTG AGTTCGGAG	GTGCCCCCAGAGAACACTATcaaacattccccattcaagacctgatcaaatgcactctactctc agccccagtcacagatctctctccagagctcagtgaaatgtttagatagaccgaagtcaca agtccttccccacagatataagtcaaatgtcccccttctccagaacctgaagtcttgccagacagtc atctctccagacctcagccagtgacctctctccagaacctcagccagacaacacctctctccaga cctcagccacagacctctctctccagaactcattcagagaaaccttccccagccccctggtcaga tgccattctccagacctcagccatacaaccttctccagacctcagccatacaaccttctct ttagacctcagccagacaacacctctctccagaactcagtcagacaacaccttctccagccccctgg tcagatgcccccttctccagacctcagccatacaacc[c/a]tctcttagaccttcagccagaca aacctctctCCAGAACTCAGCCATATGA
F5u5	F5	A	G	K	K	cds	GE172	TTTAAGAAAAT ACAGGTCTCAG CAT	TTTCTCCCATG ATTCTGTATT GT	TTTAAGAAAATACAGGTCTCAGCATtttgataaattctcaaaccaaaattggaaaacattataaga aagttatgtacacacagtagaagatgagtgctcttcaacaaacatacagtgaaatcccaatatgaa[a/g]gaagatggattttgggtctctattatcagagccaggtcagagacacactcaaatgaagta aCACAATACAGATCATCGGAGAA
F5u6	F5	G	A	E	E	cds	GE174	TCTATGCGTCT GTTCTTGTAAC	CAACCACAGGA ATGAAAACTG	TCTATGCGTCTGTCTTGTATGACagtaactactgttttggctccagagggcagcagacatcgaa cagcaggctgtgtttgctgtgttttgatgagaacaaaagctggtaaccttgaggacaacatcaacaa gttttgtgaaaatcctgatga[g/a]gtgaaacgtgatgacccccagttttatgaatcaaacatc atgacagtaagtcacagagtaactattttgttctcatCAGTTTTTCTCTGTTGGTTG
F5u7	F5	C	A	T	T	cds	GE175	TTTACACTTT CAGCTATCAAT GG	GCTTCTCTGT GAGTGTCCAG	TTTACACTTTTCAAGCTATCAATGGctaagtgcctggagacataactactcttggattctgtcttg atgacactgtccagtgccactctctgtagtggtgggacccagaaatgaaatttttgccatccacttc actggcacactctatctatgaaagagcatgagacacacttgac[c/a]ctcttccccatgc gtggagaatctgtgacgggtcaaatggataatgttggtagtaagatCTGGACACTCACAGAGG AAGC

FIG. 5EE

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
F5u8	F5	G	T	-	-	noncoding	GE174	TCTATGCGTGT GTTCCTGTACC	CAACCCAGGA ATGAAAAACTG	TCATATGGCTGTTCTTCTGACAgactatactgttt[g/t]tcctccagaggcagcagacat cgaaacagcagctgtgttgctgtgttgtagagaacaagaagctgggtacctgtgaggaacaacatca acaagtgttggaatatctgatgagtgtaacgtgtagcccccaagtttatgaattcaaacatc atgagcagtaaglcagagactatatttgttcatCAGTTTTTCATTCTCTGTGGTGTG
F5u9	F5	A	T	Y	F	cds	GE32	TCTACTAGAAG TCTGAAGATAT GAGAG	TGCCAAATTAC CCATAGAAAT	TCTACTAGAAGTCTGAAGATATGAGAGactatcttttttgtaattgtagaaaatagatatgaat atgcctttcaaataggttttgtattttatttccagtttgaatcttcttctaactctcttttaaga aaattgtctacagagagatgaaccat[a/t]ttttaagaagaacaacaacatctaccatttc aggaagcctgaatatatacttttttaatttttaagataaattTCTATGGGTAAATTGGCA
F7d10	F7	G	A	-	-	noncoding	GE293	ACCTTGAGG CAGAGAAC	AAAACCTCCT GGTGGATG	ACCTTGAGGCAGAGAACtttgcccgtcagtcctccatggggaatgcaaacagcaggggcagcac tgcagagatttcatcatggtctccagggcctcaggtctctctgcttctgcttgggttccaggg ctgcttggtcaggtgcgtcc[g/a]gggaggttttctccataaacttgggtgaaggcagtg99 gcaatccagagccagccgggcttcccaaaccccgcttgcctcggacacccccCATCCACA GGAGGGTTTT
F7d11	F7	C	T	D	D	cds	GE354	GCAGAACACCA CTGCTGACC	CGTCTTTTGC CAGTAAGATAA TCC	GCAGAACCACTGCTGACCcaggggcatggccacccccgggggtgggtctcgctgacccccag aagccctctcaggggtgcccttctgtctccagacaaggatgacagctgatatgtgtgaacg agaacggggctgtgagcagtactgagtagacacacagggcaccacagcgctctgtcgtgtgccac gggggtactctctgtggcaga[c/t]gggtgtctcgacacccacacacaggtgacaggtctcat gtccagttccagatgacacagctccctgtcccactaGGATTATCTTACTGGACAAAAGACG
F7d12	F7	G	A	-	-	noncoding	GE412	CCCTGCAGACC TAGAAATGG	CCCCATTAAT GCAGAAGAATA	CCCTGCAGACCTAGAAATGGccacagccccctccccatgcacacgggggtgaggtggcaggtggg gaaggggctga[g/a]gggggtcttctctcagggcgacacagctcagcgagcagcagcagcagc gatgagcagagcgggggtggcgaggtcatatccccagcagctacgtccccgggcaccccaa ccaagacatgcgtctgcctgcctgcacacgcgtggtctctcactgacctatgtgtgccccctet gcctcccgaaacggacgttctctgagaggacgtggcctctgtgcgtctctcatgtgtcagcggc tgggggcagctgctgacgtggcgccacggscctggagctcatggtctcaacgtgccccggct gatgacccagactgcctgcagcagtcacgggaagtgggagactccccaaatatcacggagtaca tgttctgtgcggctactcggatggcagcaaggactcctgcgaaggggagacagtgaggccccacat gccaccactacggggcagctgtgtacctgacgggactcagctgggtggcggcaggtctgcgcac cgtggggcacttgggggtgtacacacgggtctccagtatcatcgagtggtgcgaaggctcatgc gctcagagccacggcaggtcctcctgcgagcccccttctcctagcagcagcctggcctg tggagagaaggccaaaggctgcgtcgaactgtcttggcaccacaaatccccatatATTCTTCTGCAGTT AATGGG
F7u1	F7	C	T	H	H	cds	GE354	GCAGAACACCA CTGCTGACC	CGTCTTTTGC CAGTAAGATAA TCC	GCAGAACCACTGCTGACCcaggggcatggccacccccgggggtgggtctcgctgacccccag aagccctctcaggggtgcccttctgtctccagacaaggatgacagctgatatgtgtgaacg agaacggcggtgtgagcagtactgagtagacacacgggcacacagcgctctgtcgtgtgccal c[t]gaggggtactctctgctggcagcgggtgtctgcacacccacacaggtgacaggtctcat gtccagltcccagatgacacagctcctgtcccactaGGATTATCTTACTGGACAAAAGACG
F7u2	F7	G	T	G	G	cds	GE296	TGTTCTGAAT CTTTCCTAGTG G	CAGAGCTGTG TTACATTTCAA	TGTTCTGAATCTTCTCTAGTGGcaogtctcatcctccacaaatctctgcatcttcttgactttt9 ttttacacagttgaatatccatgtggaaaaataactattctcagaaaaaagaaatgccagcaaac ccaaggccgaattgtggggggcaagtggtgccccaaagg[g/t]gaggtccatggcaggttaagg cttccctggcttcaggattcccaagccctgagggtcttgagcccttTTGAATGTGAACAACAGCT CTG

FIG. 5FF

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
F7u3	F7	G	T	G	V	cds	GE412	CCCTGCAGACC TAGAAATGG	CCCCATTAACT GCAGAAGAATA	CCCTGCAGACCTAGAAATGGccacagccccatccccatgcaccagggggtgaggtggcagggtggtg gaaaggccctgaggggggttcttcttccaggcgagcagacctcagcgagcagcagcgaggtg agcagagccgggggtggcgagggtcatcatcccagcagtcagtcctccgggacacacacacac gacatcgcgctccgctcgacacgcccgtggcttctactgacctggtggtgccccctctgct gccgaacggagcttctctgagaggagcgtggcttctgctgacctggtggtgccccctgctg gccagctgctggaccgtggcgccacggccctggagctcatggctcctcaactgccccgctgctg accaggactgctgcagagtcacggaaaggagg [g/t] agactcccccaaatatcacggagttaca tgcttctgctgggtactcgatggcagaaggactctcgaaggggacagtcagtgaggcccccat gccacccactaccgggggacgtggctacactgacgggcatctcagctgggcccagggtgcgcac cgtgggccaatttgggggtacacacaggtctccagtaactgagtggtgcgaaagctcatgc gctcagagccacgcccaggagctcctctgcgagccccatctccctagccccagccccctggcctg tggagagaaaggcgaaggctcgctcgactgtctctggcaccacaaatccccataTATTCTTCTGCAGTT AATGGGG	787
F7u4	F7	C	A	D	E	cds	GE412	CCCTGCAGACC TAGAAATGG	CCCCATTAACT GCAGAAGAATA	CCCTGCAGACCTAGAAATGGccacagccccatccccatgcaccagggggtgaggtggcagggtggtg gaaaggccctgaggggggttcttcttccaggcgagcagacctcagcgagcagcagcgaggtg agcagagccgggggtggcgagggtcatcatcccagcagtcagtcctccgggacacacacacac gacatcgcgctccgctcgacacgcccgtggcttctactgacctggtggtgccccctctgct gccgaacggagcttctctgagaggagcgtggcttctgctgacctggtggtgccccctgctg gccagctgctggaccgtggcgccacggccctggagctcatggctcctcaactgccccgctgctg accaggactgctgcagagtcacggaaaggagg [c/a] tccctgaagggggacagtcgaggccccacat ctgtgcccgtactcggatggcagaaagg [c/a] tccctgaagggggacagtcgaggccccacat gccacccactaccgggggacgtggctacactgacgggcatctcagctgggcccagggtgcgcac cgtgggccaatttgggggtacacacaggtctccacaggtctccacagtcagtggtgcgaaagctcatgc gctcagagccacgcccaggagctcctctgcgagccccatctccctagccccagccccctggcctg tggagagaaaggcgaaggctcgctcgactgtctctggcaccacaaatccccataTATTCTTCTGCAGTT AATGGGG	787
F7u5	F7	C	T	T	T	cds	GE412	CCCTGCAGACC TAGAAATGG	CCCCATTAACT GCAGAAGAATA	CCCTGCAGACCTAGAAATGGccacagccccatccccatgcaccagggggtgaggtggcagggtggtg gaaaggccctgaggggggttcttcttccaggcgagcagacctcagcgagcagcagcgaggtg agcagagccgggggtggcgagggtcatcatcccagcagtcagtcctccgggac [c/t] accaa ccacgacatcgctcgctccgctcgacacgcccgtggcttctactgacctggtggtgccccctct gctgcccgaacggagcttctctgagaggagcgtggcttctgctgacctggtggttctctatggtcagcggc tggggccagctgctggaccgtggcgccacggccctggagctcatggctcctcaactgccccgct gatgacccaggactgctgcagcagtcacggaaagggtggggagactcccccaaatatcacggagttaca tgttctgtgcgggtactcggatggcagaagactcctgcaaggggagcagtcgagggccccacat gccacccactaccgggggacgtggctacactgacgggcatctcagctgggcccagggtgcgcac cgtgggccaatttgggggtacacacaggtctccacaggtctccacagtcagtggtgcgaaagctcatgc gctcagagccacgcccaggagctcctctgcgagccccatctccctagccccagccccctggcctg tggagagaaaggcgaaggctcgctcgactgtctctggcaccacaaatccccataTATTCTTCTGCAGTT AATGGGG	787

FIG. 5GG

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
F7u6	F7	A	T	E	V	cds	GE412	CCCTGCAGACC TAGAAATGG	CCCATTAAC GCAGAAGAATA	CCCTGCAGACCTAGAAATGGccacagccccatccccatgcacacaggggtgaggtggcaggtgggtg gaaagggcctgagggggggtctctctccagggagacacccctcagcgagcagcagcggggatg agcagagccggcggtggcgaggtcctccatccacagcagcagctacgtccgggacacacacacac gacatcgcgctgctccgctgacacacccggtggtcctcactgacacacacacacacacacac gcccgaacggagctctctg[a/t]gagacgctggcctctgctgctctctcattggtcagcggc tggggcagctgctggacgctggcgccacggccctggagctcattggtcctcaacgtgccccgct gatgaaccaggaactgctgagcagctcagcagctcagcaggtggagagctcccccaaatatcacaggagtaca tgctctgctggcgtactcgtatggcagcaaggactcctgcagggggagctggagggccacacat gccacccactacgggggacgtggtaactgacgggcatcgctcagctggggccaggggtgcgcaac cgtgggcccacttgggggtgacacacaggggtctccagctacatcgagtggtgcaaaagctcatgc gctcagagccacgccccaggtcctctgagagccccatttccctagccccagcagccccggcctg tggagagaaagccaaaggctgctgagactgctctggcaccacaaatccccatATACTTCTGTCAGTT AATGGGG
F7u7	F7	C	T	A	A	cds	GE412	CCCTGCAGACC TAGAAATGG	CCCATTAAC GCAGAAGAATA	CCCTGCAGACCTAGAAATGGccacagccccatccccatgcacacaggggtgaggtggcaggtgggtg gaaagggcctgagggggggtctctctccagggagcagcagcctcagcgagcagcagcggggatg agcagagccggcggtggcgaggtcctccatccacagcagcagctacgtccgggacacacacacac gacatcgcgctgctccgctgacacacccggtggtcctcactgacacacacacacacacacac gcccgaacggagctctctgagaggaagctggcctctgctgctctctcattggtcagcggcctggg gcccagctgctggacgctggcgccacggccctggagctcattggtcctcaacgtgccccgctgatg accaggaactgctgagcagctcagcagaggtgggagactcccccaaatatcacaggagtacatgtt ctgtgc[t]ggctactcgtatggcagcaggaactcctgcaggggggagcagctggagggccacacat gccacccactacgggggacgtggtaactgacgggcatcgctcagctggggccagggctgcgcaac cgtgggcccacttgggggtgacacacaggggtctccagctacatcgagtggtgcaaaagctcatgc gctcagagccacgccccaggtcctctgagagccccatttccctagccccagcagccccggcctg tggagagaaagccaaaggctgctgagactgctctggcaccacaaatccccatATACTTCTGTCAGTT AATGGGG
F7u8	F7	T	A	V	D	cds	GE412	CCCTGCAGACC TAGAAATGG	CCCATTAAC GCAGAAGAATA	CCCTGCAGACCTAGAAATGGccacagccccatccccatgcacacaggggtgaggtggcaggtgggtg gaaagggcctgagggggggtctctctccagggagcagcagcctcagcgagcagcagcggggatg agcagagccggcggtggcgaggtcctccatccacagcagcagctacgtccgggacacacacacaa ccacgacatcgctgctccgctgacacacccggtggtcctcactgacacacacacacacacacac gcccgaacggagctctctgagaggaagctggcctctgctgctctctcattggtcagcggc tggggcagctgctggacgctggcgccacggccctggagctcattggtcctcaacgtgccccgct gatgaaccaggaactgctgagcagctcagcagaggtgggagactcccccaaatatcacaggagtaca tgctctgctggcgtactcgtatggcagcaaggactcctgcaggggggagcagctggagggccacacat gccacccactacgggggacgtggtaactgacgggcatcgctcagctggggccagggctgcgcaac cgtgggcccacttgggggtgacacacaggggtctccagctacatcgagtggtgcaaaagctcatgc gctcagagccacgccccaggtcctctgagagccccatttccctagccccagcagccccggcctg tggagagaaagccaaaggctgctgagactgctctggcaccacaaatccccatATACTTCTGTCAGTT AATGGGG

FIG. 5HHH

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
F7u9	F7	G	A	R	Q	cds	GE412	CCCTGCAGACCTAGAAATGGccacagcccatcccatgcacagggggtgaggtggcaggtgggtg	CCCAATTAACTGCAGAGAAATA	CCCTGCAGACCTAGAAATGGccacagcccatcccatgcacagggggtgaggtggcaggtgggtg gaaagggcctgaggggggtggtctctctccagggagcagcctcagcagagcagcagcagggggtg agcagagccgggggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtg gacatcggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtg gcccgaacgggacgttctctgagggagcgtggtggtggtggtggtggtggtggtggtggtggtg gcccagctggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtg accagagctgctgagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc ctgtgcccgtactcgtgagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc cccactacc [g/a]gggcacgtggtggtggtggtggtggtggtggtggtggtggtggtggtggtg cgtggcccacttttgggggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtg gctcagagccacgcccaggggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtg tgagagagaaagccaggggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtg AATGGGG	787
F9d8	F9	G	T	-	-	noncoding	GE138	TTCAGATGCAGAGCATAGAATA	TGGACTGATCTTTCTGAGTCCT	TTCAGATGCAGAGCATAGAATAcctttaaagaacacttctctttaaataattttaaagcat ccatatatttattgtat [g/t]ttaaaggttataaaagataagaaatcaatataccaaacacactt agatatattaccgttatttcttcttcttcttcttcttcttcttcttcttcttcttcttcttctt taagcaattcatttc tgcatataatagataatataataacttctctctctctctctctctctctctctctctctctctc GGCTCAATCTCAATTTTGTAAATacatggttcccatctgccaatgagaaatatacaggttactaat ttc accctggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtg tttggataacatacctcaagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc gatgccaacacaggtcaattctctctgaggtggtggtggtggtggtggtggtggtggtggtggtg cagctggcagaagacacagggccaggtgggagcagctgaggtggtggtggtggtggtggtggtggtg TTTTGCCCTATTCTCTGTAACCCAGcacacacacacacacacacacacacacacacacacacac agagctcttaatttttct gctctatcgtttaaagaaataatggtggtggtggtggtggtggtggtggtggtggtggtggtggtg acagttgtcgcaggttaataacacagaaagaaataataatctgcagcagcagcagcagcagcagcag attggtacaccataatttct GGCTCAATCTCAATTTTGTAAATacatggttcccatctgccaatgagaaatatacaggttactaat ttc accctggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtg ggataacatcactcaaaagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc gatgccaacacaggtcaattctctctgaggtggtggtggtggtggtggtggtggtggtggtggtg cagctggcagaagacacagggccaggtgggagcagctgaggtggtggtggtggtggtggtggtggtg CAATGAGTATCTACAGGGGAGGAGccgggcatctctagcaggttctagcaggttctagcaggttct ccatctcctcaagatgagatcaggtggtggtggtggtggtggtggtggtggtggtggtggtggtg gacatttaattct gt [a/g]actatttttttgaatactcactcaggttctctctctctctctctctctctctctctctct AATGCAATATTGGTG	316
F9u1	F9	G	A	A	T	cds	GE164	GGCTCAATCTCAATTTTGTAAATacatggttcccatctgccaatgagaaatatacaggttactaat CAATTTTGTAAAT	AATAGCCTCAGTCTCCACCT	GGCTCAATCTCAATTTTGTAAATacatggttcccatctgccaatgagaaatatacaggttactaat ttc accctggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtg tttggataacatacctcaagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc gatgccaacacaggtcaattctctctgaggtggtggtggtggtggtggtggtggtggtggtggtg cagctggcagaagacacagggccaggtgggagcagctgaggtggtggtggtggtggtggtggtggtg TTTTGCCCTATTCTCTGTAACCCAGcacacacacacacacacacacacacacacacacacacac agagctcttaatttttct gctctatcgtttaaagaaataatggtggtggtggtggtggtggtggtggtggtggtggtggtggtg acagttgtcgcaggttaataacacagaaagaaataataatctgcagcagcagcagcagcagcagcag attggtacaccataatttct GGCTCAATCTCAATTTTGTAAATacatggttcccatctgccaatgagaaatatacaggttactaat ttc accctggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtg ggataacatcactcaaaagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc gatgccaacacaggtcaattctctctgaggtggtggtggtggtggtggtggtggtggtggtggtg cagctggcagaagacacagggccaggtgggagcagctgaggtggtggtggtggtggtggtggtggtg CAATGAGTATCTACAGGGGAGGAGccgggcatctctagcaggttctagcaggttctagcaggttct ccatctcctcaagatgagatcaggtggtggtggtggtggtggtggtggtggtggtggtggtggtg gacatttaattct gt [a/g]actatttttttgaatactcactcaggttctctctctctctctctctctctctctctctct AATGCAATATTGGTG	366
F9u2	F9	A	T	K	N	cds	GE142	TTTTGCCCTATTCTCTGTAACCCAG	GGCCTCAATCTCAATTTTGTAAAT	TTTTGCCCTATTCTCTGTAACCCAGcacacacacacacacacacacacacacacacacacacac agagctcttaatttttct gctctatcgtttaaagaaataatggtggtggtggtggtggtggtggtggtggtggtggtggtggtg acagttgtcgcaggttaataacacagaaagaaataataatctgcagcagcagcagcagcagcagcag attggtacaccataatttct GGCCTCAATCTCAATTTTGTAAATacatggttcccatctgccaatgagaaatatacaggttactaat ttc accctggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtg ggataacatcactcaaaagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc gatgccaacacaggtcaattctctctgaggtggtggtggtggtggtggtggtggtggtggtggtg cagctggcagaagacacagggccaggtgggagcagctgaggtggtggtggtggtggtggtggtggtg CAATGAGTATCTACAGGGGAGGAGccgggcatctctagcaggttctagcaggttctagcaggttct ccatctcctcaagatgagatcaggtggtggtggtggtggtggtggtggtggtggtggtggtggtg gacatttaattct gt [a/g]actatttttttgaatactcactcaggttctctctctctctctctctctctctctctctct AATGCAATATTGGTG	316
F9u3	F9	G	T	R	R	cds	GE164	GGCCTCAATCTCAATTTTGTAAAT	AATAGCCTCAGTCTCCACCT	GGCCTCAATCTCAATTTTGTAAATacatggttcccatctgccaatgagaaatatacaggttactaat ttc accctggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtggtg ggataacatcactcaaaagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc gatgccaacacaggtcaattctctctgaggtggtggtggtggtggtggtggtggtggtggtggtg cagctggcagaagacacagggccaggtgggagcagctgaggtggtggtggtggtggtggtggtggtg CAATGAGTATCTACAGGGGAGGAGccgggcatctctagcaggttctagcaggttctagcaggttct ccatctcctcaagatgagatcaggtggtggtggtggtggtggtggtggtggtggtggtggtggtg gacatttaattct gt [a/g]actatttttttgaatactcactcaggttctctctctctctctctctctctctctctctct AATGCAATATTGGTG	366
F9u4	F9	A	G	-	-	noncoding	GE103	CAATGAGTATCTACAGGGGAGGAG	CACCAATATTG CATTTCACAGT	CAATGAGTATCTACAGGGGAGGAGccgggcatctctagcaggttctagcaggttctagcaggttct ccatctcctcaagatgagatcaggtggtggtggtggtggtggtggtggtggtggtggtggtggtg gacatttaattct gt [a/g]actatttttttgaatactcactcaggttctctctctctctctctctctctctctctctct AATGCAATATTGGTG	275

FIG. 5III

[illegible]

FIG. 5JJJ

68/178

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
FGA10	FGA	G	C	A	P	cds	GE414	GAGGCTTAACA ACCCAGACT	AAAAAGTGTAG TTTCAATGACG TGTA	GAGGCTTAACAACCCAGACTGGGGCACATTGAGAGGTTGTcaggaaatgtaagtccaggggacaa 821 ggagagagtaccacacagaaactgggtcaccttctaagagagataaagagctcaggactggtaaa ggaaaggtcacctctggtagacacacacacacgctggttcacgtcttaaaacccgttactaagac tggtattggctcctgaggtcacaaagagttaccacaaagtggtgacctccgaagatggttctg actgtcccgagggaatggatttaggcacattgtctggcataggtactctggatgggtccgcat aggcacctgatgaa[g/c]ctgctctctcgacacctgcctcaactggaaacacattcccagggt tcttctacactatgttagggaggtttgtcagtgagctgagtcaggggctcagaatctggcatc ttcacaaatacaaaagggaatccagttctcacacccctgggagatagtgaaattcccttccgtggtaa atcttcaagttacagcaaaatctactagtagcagagttacacagagagagactccacattg aaagcaagagctataaaatggcagatgagccgggaagtgaagcagatcatgaaggaacacatagc accaagagagggccatgctaaatctcgcctgtcagaggtatccacacttctcttggggaagcc ttccctgtccctcagactaagttaaatattctgcacagtggtcccatggcccttgcatttcc ttcttaactctctgtTACACGTCATTTGAACACTACACTTTT
FGA11	FGA	G	A	G	E	cds	GE415	TTAACTACCAG GAACCTCAATAG ACG	CTGACACCTCT TCAAATGTGC	TTAACTACCAGGAACCTCAATAGAGTgttattgtattgtatcacatttctcttttattttt 825 tccctctctctaggtggacattgatatattagatccgatcttgcagaggtcatgcagtagggct ttagctcgtgaagttagtctgaaggactatgaagatcagcagaagcaactgaaacaggcoattgc caaagacttacttccctctagataggaacacacttaccactgtataaaatgaacacagttccag acttggttcccggaattttaagagccaggttcagaaggtacccacagatgggaaggtcattaca gacatgccagatgagaatggatttagagacacctggttggaatgagattactcagagaggtc cactcttattggaacccggtacagagacggaagcccggaacccctgagctctggagaggtgga acttggaacccctgggaactctggaactctggaaccccggaacccctggaactctggagagctc tggaactgggaactctggaactctggaactctggaactctggaactctggaactctggaactc atcttggaactctggaactctggaactctggaactctggaactctggaactctggaactctgga tagtactggacaaatggcactctggaactctggaactctggaactctggaactctggaactctgga cgaggcctaacacacccagactggggcactTTTGAAGAGGTGTCTAG
FGA12	FGA	T	A	S	T	cds	GE415	TTAACTACCAG GAACCTCAATAG ACG	CTGACACCTCT TCAAATGTGC	TTAACTACCAGGAACCTCAATAGAGTgttattgtattgtatcacatttctcttttattttt 825 tccctctctctaggtggacattgatatattagatccgatcttgcagaggtcatgcagtagggct ttagctcgtgaagttagtctgaaggactatgaagatcagcagaagcaactgaaacaggcoattgc caaagacttacttccctctagataggaacacacttaccactgtataaaatgaacacagttccag acttggttcccggaattttaagagccaggttcagaaggtacccacagatgggaaggtcattaca gacatgccagatgagaatggatttagagacacctggttggaatgagattactcagagaggtc cactcttattggaacccggtacagagacggaagcccggaacccctgagctctggagaggtgga acttggaactctggaactctggaactctggaactctggaactctggaactctggaactctgga actgcaacccctgggaactctggaactctggaactctggaactctggaactctggaactctgga tggaactgggaactctggaactctggaactctggaactctggaactctggaactctggaactc atcttggaactctggaactctggaactctggaactctggaactctggaactctggaactctgga tagtactggacaaatggcactctggaactctggaactctggaactctggaactctggaactctgga cgaggcctaacacacccagactggggcactTTTGAAGAGGTGTCTAG

FIG. 5KKK

[illegible]

FIG. 5LLL

[illegible]

FIG. 5MM

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
FGAu8	FGA	T	C	P	P	cds	GE414	GAGGCCTAACAA ACCCAGACT	AAAAAGTGCTAG TTTCAATGACG TGTA	GAGGCCTAACAAACCCAGACTGGggccacatttgaagagggtgtcaggaaagttaagtccagggaacaa ggagagagtaccacacagaaactgggtcactctaaagagagataaagagctcaggactcggtaaa gagaaggtcacctctgttagcacacaacacacacggtgtgtcactgtctaaacacgttactaaagac tgttatgggtcctgattggttcacaaagaagttaccacaaagaagtggtgactccgaagaaggtctctg actgtcccgagggaatggatttaggcacattgtctggcataggtactctgtgactcgaaggttccgccat aggcaccctt/cjgataagctgctctctcgacacactcctcaactggaacacacattccacaggtt tcttccactatgttaggagagttgtcagtgagactgagcttaggggctcagaaatctggccatc ttcacaaatacaaaaggaaatccagttctcactcaccctggatagctgaattccctccctgggttaa atcttcaagttacagcaaaacatttactagtagcacaggtttacaacagagaggagactccacatttg aaagcaagagctataaaatggcagatgagggcgggaagtggaacgcatcatgaaggaaacacatagc accagagggggccatgctaaatctgcgcctgtcagaggtatccacacttctctcttgggaagcc ttccctgtccccctagactaagttaaatatttctgcacaggttcccatggtcccttgcatttccc ttcttaactctctgtttACAGCTCAATTGAAACTACACTTTT	1821
FGAu9	FGA	G	C	S	T	cds	GE415	TTAACTACCCAG GAACTCAATAG ACG	CTGACACCTCT TCAATGTGC	TTAACTACCCAGGAACTCAAATAGACGGtagtttatgtattgtatctacatttctcttatttcttct tccccctctctaggtgacattgatataagatccgatcttctgtcgagggtcatgcagtagggct ttagctgtgaagtagatctgaaggactatgaagatcagcagaagcacttgaacaggtcatctgc caagacttacttcccccgaaalttaagggccagcttcagaggttaccgccagagtggaaggcattaaca acttggttcccccgaaalttaagggccagcttcagaggttaccgccagagtggaaggcattaaca gacatgcgcagatgagatggagttagagagacacctgggtggaaatagatctactcgaggaggtctc cacctctatggaaacggatcagagacggaaagcccccagaaacccctggggactctgggaactggagg actctgggagctctggacctggagttactggaaaccccccagaaacccctggggactctgggaactggagg actgcaacctggaaacctgggagctctggacctggagactactggaa[g/c]ctggaaactctggga gctctggaactggagtagtggaaacaaacccctggagcccttagactggttagtaccggaaacc tggaatctctggcagctctgaacgcgggaagtgctggggcactggacctctgagagctctgtctctgg tagtactggacaatggcactctgaaatctggaaagttttagggcagatagccccaggtctctgggaacg cgaggcctaacaacccagactggggCACATTTGAAGAGGTGTCTAC	1825
FGBd12	FGB	T	A	-	-	noncoding	GE336	GTAACCAATTC TGAAGTCATTC CT	CAATTCATTT CATAACTATAA GCAA	GTAACCAATTCGAAAGTCATTCCTAGcagagaggac[t/a]cagatatatatataggattgaagatctctcaa tcaagttaaagctctacatgaagaaaggaaggttctctggagcttccacaactttaaaccactgaacaa tctattattgtctactatgtgtgttttcttagttaagttcccaaggttgcacagacaatgagaggagg tgaattttttaaagcattattattatttagtagtagtattattataagatgtaacataatcata ttatgtgcttatttttaagaaatttagcattgctttATAGTTATGAAATGGAATTTG	314
FGBd13	FGB	A	G	K	E	cds	GE336	GTAACCAATTC TGAAGTCATTC CT	CAATTCATTT CATAACTATAA GCAA	GTAACCAATTCGAAAGTCATTCCTAGcagagaggactcagatatatataggattgaagatctctcaa gttaagctctacatgaaaggaaggttctctggagcttccacaactttaaaccactgaacaa tctattattgtctactatgtgtgttttcttagttaagttcccaaggttgcacagacaatgagaggagg tgaattttttaaagcattattattatttagtagtagtattattataagatgtaacataatcata ttatgtgcttatttttaagaaatttagcattgctttATAGTTATGAAATGGAATTTG	314
FGBd14	FGB	T	A	-	-	noncoding	GE336	GTAACCAATTC TGAAGTCATTC CT	CAATTCATTT CATAACTATAA GCAA	GTAACCAATTCGAAAGTCATTCCTAGcagagaggactcagatatatataggattgaagatctctcaa gttaagctctacatgaaaggaaggttctctggagcttccacaactttaaaccactgaacatcta ttattgtctactatgtgtgttttcttagttaagttcccaaggttgcacagacaatgagagggtgaa ttttttaaagcattattata[t/a]tatttagtagtattattataagatgtaacataatcata ttatgtgcttatttttaagaaatttagcattgctttATAGTTATGAAATGGAATTTG	314

FIG. 5NNN

73/178

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
FGBu2	FGB	A	T	E	V	cds	GE390	TCATAACTGCTTGGTGTAGCT	CCACTTAGCATTTTGTGTGTG	TCATAACTGCTTGGTGTAGCTCAGTGTtaaatagtttattctcagaaaaatcaaaatgttatagttaaatacattagtttctaggggaaaaactcctcagaaactgtaactattctacatacaatttccacagataatgla/tjaaatgtagtcaactcctcagaaactggaagagcaaaattatataatagatgagactgtgtaataagcaatatacccaactaaccttctgtgtctcgttcaatccctggaagaaactgagaa gcaaaatcaaaaagttagaatctgtctcagctcaaatggaatattgtcgcaccccatgcactgtcagltgcaaatattcctgtgtgtgtgcaaaaggttaactgattcataaacaatttttagagagttccagaaagaaactcacacaccccaaaataagagaacaaacaCAACAACAAATGCTAAGTGG	452
FGBu3	FGB	G	T	V	V	cds	GE330	GGGATTCAGATATTATTTTCAAGTGCACAT	GCTTCCAACAA TGAATGTTTTT	GGGATTCAGATATTATTTTCAAGTGCACATatttctgtgtgtgtgttaatatatgctctcttttgtttctgtcaacacaaagagatggacagt[ig/t]attcagaaccgtcaagacggtagtgtgtgacttttggcaggaaatgggataccatatataaacacaggat[ig/t]tggaataattgttgcacacacacagatgggaagaattacgtggcctaccaggtaacgaacaggcatgcaaaaataaaatcattctatttgaatgggattttttttaaattaaaaaacatttcttgggaagc	289
FGBu4	FGB	G	A	R	K	cds	GE382	ATGGGTAATCTGCAAAACGTA	TCAAAAAGTCA CACTCAGCTCTG	ATGGGTAATCTGCAAAACGTAacttgaccacccgtagttctgtttctataaacgccaaacacattttctttcaggttaacatcagatcccagaaaaacagtttctaaagaagacggttgggtggaagggtggtataatagatgtcatgtagcagcaatcccaacggcagatactactgggtggacagtaacacctgggacalggcaagcatgagcagatgtagttagttagtgaatggaaagggtcactggtactcaatga[ig/a]gaagatgagtagaagatcagggccttctccacacagcaaatagttcccaatacagtagattttgtctctgtatgtgacaacattttgtacattattgttgaattttcttcttcacattatctctctctctctctcaagCAGACGTCAGTGTGACTTTTTGA	435
FGBu5	FGB	T	A	G	G	cds	GE477	GAATAGTTACATTCCTCAATCTTCTA	TGACTACAGGC TTTCTCTGCAT	GAATAGTTACATTCCTCAATCTTCTAaacactctgtattatatttctgctctcattctcctgttagggttctctcagtgccctgg[ig/a]catgcaccccttgacaagaagagagagaggtcctccagcctgaggcctgccccacccatcagtgagggtggctcgtcctccagcctcagcagcaagcagctgcccaccaaaaagaaagttagaaagaaaagccctgtagtgcagggtgtctctcagctgacccagacctgggtggcactgagttctctgcagtggtggctctctcATGCAGAGAAAGCCTGTAGTCA	319
FGBu6	FGB	A	T	A	A	cds	GE330	GGGATTCAGATATTATTTTCAAGTGCACAT	GCTTCCAACAA TGAATGTTTTT	GGGATTCAGATATTATTTTCAAGTGCACATatttctgtgtgtgttaatatatgctctcttttgtttctgtcaacacaaagagatggacagtgtatcagaacccgtcaagcggtagtgtgacttttggcaggaaatgggataccatatataaacacaggat[ig/t]tggaataattgtgc[ig/a/t]accaacacagatgggaagaattactgtggcctaccaggtaacgaacaggcatgcaaaaataaaatcattctatttgaatgggattttttttaaattaaaaaacatttcttgggaagc	289
FGBu7	FGB	A	G	R	R	cds	GE477	GAATAGTTACATTCCTCAATCTTCTA	TGACTACAGGC TTTCTCTGCAT	GAATAGTTACATTCCTCAATCTTCTAaacactctgtattatatttctgctctcattctcctgttagggttctctcagtgccctgg[ig/a/g]ccccttgacaagaagagagagaggtcctccagcctgaggcctgccccacccatcagtgagggtggctcgtcctccagcctcagcagcaagcagctgcccactcaaaaagaaagttagaaagaaaagccctgtagtgcagggtgtctcagctgacccagacctgggtgggtgcaactgagttctctgcagtggtggctctctcATGCAGAGAAAGCCTGTAGTCA	319
FGBu8	FGB	C	T	H	H	cds	GE477	GAATAGTTACATTCCTCAATCTTCTA	TGACTACAGGC TTTCTCTGCAT	GAATAGTTACATTCCTCAATCTTCTAaacactctgtattatatttctgctctcattctcctgttagggttctctcagtgccctgg[ig/a]ccccttgacaagaagagagagaggtcctccagcctgaggcctgccccacccatcagtgagggtggctcgtcctccagcctcagcagcaagcagctgcccactcaaaaagtagaaagaaaagccctgtagtgcagggtgtcttca[ig/t]gctgacccagacctgggtgggtgcaactgagttctctgcagtggtggctctctcATGCAGAGAAAGCCTGTAGTCA	319

FIG. 5PPP

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
FFGBu9	FGB	C	T	Y	Y	cds	GE392	AAGGGAAGAAA GGCAGTTTT	CCGAGGAAGTG GTAGCTATTAA A	AAGGGAAGAAAAGGCAGTTTTTtagtttccccaaaatttttatttttggtagagagatttttatttttggttt ttcttttaggtgaatttggcttggaattgataaaattagccagcttaccaggtaccaggtggaagccacac gaacttttgatagaaattggaggactggaagagacaaagtaaaaggtcacatgtagagatttcac tgtacgaattgaagccaaataaccagatctcagatgacaaata[c/t]agaggaacagccggt aatgccctaatggatggagcatctcagctgatgggggaaaaacaggaccatgaccattcacaaacgg catgtttcttcagcacgtatgacagagacaattgacgggtggtatgtgtggcactctttgtctctcggc tttaaaatcacactaatattactactcagaattcattacaataattttTAATAGCTACCACTTCC TGGG
FFGGd3	FGG	A	G	M	V	cds	GE337	TTCTGTAATAGA CAGCTCTTCAT AGACT	GCAGTTAATTT TCTACAAATCA TCC	TTCTGTAATAGACAGCTTTTCATAGACTTgcagaggtaaaagattccagaataatgatattgtlaca tctacgacttggttttaggtggcacttactcctcaaaaagcatctactcctaatggtttatgataatggca ttatttggggccacttggaataacccgggtggttattccatgaagaaaaaccact[a/g]tgaagataaat ccattcaacagactcacaaattggagaggacagcaacacccctgggggagccaaacaggtca gaccagagcacccctgcggaaacagaaatatgactcacttaccctgaggtGATTTGTAGAAAATT AACTGC
FFGGd4	FGG	A	G	-	-	noncoding	GE349	AAATACTTAG CAGTTTCCAAA GAAAA	TGGGTAGCCAC TTTCTRAACTA TTC	AAATACTTAGCAGTTTCCAAAAGAAAAtataaaaattactcttctgaagggaaataacttatttttgt cttctatttttggttacttattgtttctgttattttagatatttgcaggaataataataattcaata atcaaaagattgttaacctgaaagagaggttagccagcttgaagcacagtgccaggaaaccttgc aaagacacggtgcaaatccatgatatacactgggaaaggtaactgatgaaggtttatatgtgggattta ggttcatacaaaagtaagtaattgtaaaggagaaagtatgtactgg[a/g]aaagtataggAAATAGTTT AGAAAGTGGCTACCCA
FFGGd5	FGG	G	A	G	R	cds	GE360	TGCTGATGTGA AAAGTAAGAAA AT	CAAGGTGCTTA GAAAAGTATCT GC	TGCTGATGTGAAAAGTAAGAAAATtattcttggaaaatgaatagtttactacatglttaaaagcta tttttcaaggctggcagagcttaccctgcatttcaaacacagtaaaagtcgattctctctctct agattgtcaagacattagccaataaaggagctaaacagagc[g/a]ggctttactttattataaacct ctgaagctaacacagcaattctactgtgaatcgatgggtctggaaatggatggactgt gtttcagaaggtaatttttcccccccatgtgtatttaataaatctccatctgttttcttgccata tgGCAGATACTTTTCTTAAGCACCTTNG
FFGGd6	FGG	T	A	-	-	noncoding	GE372	GAACCAAGTGT CTGTATTTTTC AC	CCATTGCTAT TGATAGTTGGA AAG	GAACCAAGTGTCTGTATTTTTCACaaaattgttgacagcatctcttttaca[t/a]gcattgatag tctattttctcttcttctctctcttgcctcttgcataatgtgtaattagagacttgatggcagtgtagatttcaag aaaaactggattcaatataaagaaggatttgacactgtctctactggcacaacagaaatttctg gctgggaaatgagaagalttcattgtaagcacacagctctgcccataatgcatttaagagtggtg aactggagagactggaatggcagaaccaggtactgttttgaataatgacttccaaactttttattgttaa agattgctgtggaattgcaCTTTCCAACTATCAATAGACAATGG
FFGGu1	FGG	A	T	Y	F	cds	GE404	CATCTACGAA AGAGGGAACCT	TCCACTTCCAG TTTCAAGAAC T	CATCTACGAAAGAGGGAACTTctgagatccctgagggggtcagcatgtgatggttctatttcttcc ttctctcagtagctgcagact[a/t]tgccatgttcaaggtgggacctgaagctgacagtagtaccg cctaaacatatgcctactctgcctggtggggatgctggagatgccttttgatggttcttatttggcg atgatcctagtgaagaattttcacatcccatataatggcatgcagttcagtaacctgggacaatgac aatgataagtttgaaggcaactgtgtgaacaggatggtatggttgggtggaatgaacagtgatca cgctggccatctcaatggagtttataccagaggtatgtttctctttcttagatcccaagttaatg tatagtgataactatttcatataaaaaataataatagatatgaagaaatgaagaataaatttataa agatacagggatttttcatgttcttatttcaactaAGTTCTTTGAACTGGAAGTGA

FIG. 5QQQ

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')
FFGGu2	FGG	A	C	I	I	cds	GE372	GAACCAAGTGCT CTGTATTTTTG AC	CCATTTGCTAT TGATAGTTGGA AAG
FSHRu1	FSHR	A	G	T	A	cds	GE667	GCTCTGAGCTT CATCCA	AGCAAAAATCC AGCCCATCA
FSHRu2	FSHR	G	A	S	N	cds	GE648	GCCATGCTGCC AGTGTC	TGTTTTAGTTT TGGGCTAAAT
FSHRu3	FSHR	C	G	S	R	cds	GE648	GCCATGCTGCC AGTGTC	TGTTTTAGTTT TGGGCTAAAT

FIG. 5PARR

[illegible]

FIG. 5SSS

Poly Id	Gene	ref N ^o	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
FSHu2	FSH	G	T	S	I	cds	GE561	T ^T TCTCAGT ^{TT} CTAGTGGGCT ^{TT} C	GGTACCTACCC TGGTGTAACA	TTTCTCAGTTTCTAGTGGCTTCattggttgcctccagaccaggaatgaagacacactccagttttt cttcctttctgttgcgaaagcaatctgtgcaata[g/t]ctgtgagctgacacacatcaccc attgcaabagagaagaagaatgtgcttctgcataagcatcaacaccacttgggtgctggcta CTGCTACACCAGGGTAGGTACC
GABRB1 a7	GABRB1	A	G	-	-	noncoding	GE1089	CAGATGGGTATT CAAAATGAT ^{TT} CCTA	AACCATCTTCT TCCTTTCACAG	CAGATGGTATTCAAAATGAT ^{TT} TCTTaaacttggtttaaccgtgctgtttttatttggtttcagatg gctataccactgatgacattgaatttctactggaatggaggagaaggggcagtcactcgtgtttaa aaaaatcgaaacttctcaatttcaattgttgactacaagatgggtcttaagaaggtggagttcac aacagggtgaggttgtttcccccaa[a/g]atgtactaggggtgctgtaaaaggaaagagatggtt AACATGATTGGGGTAGAGAGcctggaaatgaaatgctcatcacttttgaatgtttctttttt ctctctctctatcatcagaatcacaacacagctgcattgattgattgattcttgcagaatataccac tggatgagcagaactgcacctggagat[c/a]gaagttgtgattgattgagcaggggaatga aaaagagggtatctctcttgacccagttgaATTCACTTCTCAGTGAATTAATAGCA
GABRB1 a8	GABRB1	C	A	I	I	cds	GE1271	AACATGATTG GGGCTAGGA	TGCTAATTAAAC TCAGTGAGAG TTGAAT	AACATGATTGGGGTAGAGAGcctggaaatgaaatgctcatcacttttgaatgtttctttttt ctctctctctatcatcagaatcacaacacagctgcattgattgattgattcttgcagaatataccac tggatgagcagaactgcacctggagat[c/a]gaagttgtgattgattgagcaggggaatga aaaagagggtatctctcttgacccagttgaATTCACTTCTCAGTGAATTAATAGCA
GABRB1 d3	GABRB1	G	A	-	-	noncoding	GE1035	CCAGCCTGCTG TCACTGAG	GTCCATTTCCC ATCTGGGTA	CCAGCCTGCTGCTCAGAGAGaactctgttctctaattgtggccacactcccc[g/a]gcaggggcccc ccgtgcagcttggaatggatcgaatgtccacagcatagacatggtctccgaagtgaaatggtg agtggcctccccaggggccggctcggttcggctTACGCAGATGGGAATGGAC
GABRB1 d4	GABRB1	C	T	I	I	cds	GE1089	CAGATGGGTATT CAAAATGAT ^{TT} CCTA	AACCATCTTCT TCCTTTCACAG	CAGATGGTATTCAAAATGAT ^{TT} TCTTaaacttggtttaaccgtgctgtttttatttggtttcagatg gctataccactgatgacattgaatttctactggaatggaggagaaggggcagtcactcgtgtttaa aaaat[c/t]gaacttctcaatttcaattgttgactacaagatgggtcttaagaaggtggaggt tcacaacagggtgaggttgtttcccccaaaatgtactaggggtgctgtaaaaggaaagagatggtt CTCTCAATCTTGAAAAAGGAacttaatagtggc[a/g]ccttcagctaaagtgtgtcttctctct ctcacaggaaatcacgcggtgcttaacatgacaacacacatgcagccaccctcagggagaccctgccc aaagatcccttatgtcaaaagctgtatatttatctgatgggttgccttctgttctgttctcctgg ctctgtggagtagtgccttggtaaattacatctcttcttggaaagccctcagaaaaggagct agcaaaagaagaccagagtgccaatgagaagaataaactggagatgaataaagtcacagtaagata ttaaatactcctaacaatatcttctgtttaaatttatcagcatCATATGCTTCGGGCTCTC
GABRB1 d5	GABRB1	A	G	-	-	noncoding	GE1134	CCTCTCAATCT TGAAAAAGGA	GAGAGCCCGAG GCATCAT	TGAAAAACAGGCATAAGGTCCTgcacactgtgtccgagctgttctttttggccatcaggtgcagccc cacggtacaactctctccagcacccttgaaaaatccggaatgagacgagtggtcctcggaagtgctcac gagcgtgagcagcccccaaggccaccatgtactcctatgcacgcgcagcatccagtcacccaagc ccctgagcagccgcgagccctacgggcgcgcctcctggagcagcgggtacccagcaaggggcgc atccgagggctgctcccgactcaagtcagaatcccgactt[g/a]actgattgaattcca tagacaagtggtcccgaaatgttttccccatcaccttctctttttaaattgctcgtctatggctt tactatgtacactgaggtctgttcttaatttccatttagactacttctctctctctctctctctt TAACCTTACAGGTCCCCA
GABRB1 d6	GABRB1	G	A	L	L	cds	GE1144	TGAAAAACAGGC AAAGGTCC	TGGGGACCTGT ^T AAGGTTAAAAA	TGAAAAACAGGCATAAGGTCCTgcacactgtgtccgagctgttctttttggccatcaggtgcagccc cacggtacaactctctccagcacccttgaaaaatccggaatgagacgagtggtcctcggaagtgctcac gagcgtgagcagcccccaaggccaccatgtactcctatgcacgcgcagcatccagtcacccaagc ccctgagcagccgcgagccctacgggcgcgcctcctggagcagcgggtacccagcaaggggcgc atccgagggctgctcccgactcaagtcagaatcccgactt[g/a]actgattgaattcca tagacaagtggtcccgaaatgttttccccatcaccttctctttttaaattgctcgtctatggctt tactatgtacactgaggtctgttcttaatttccatttagactacttctctctctctctctctctt TAACCTTACAGGTCCCCA
GABRB1 u1	GABRB1	G	A	T	T	cds	GE1134	CCTCTCAATCT TGAAAAAGGA	GAGAGCCCGAG GCATCAT	CCTCTCAATCTTGAAAAAGGAacttaatagtggcacacttcagctaagtgtgtctctctcttcca caggaataccagac[g/a]gtgcttaacatgacaacacatcagcaccacactcagggagaccctgccc aaagatcccttatgtcaaaagcattgattatttatctgatgggttgccttctgttctgttctcctgg ctctgtggagtagtgccttggtaaattacatcttcttctggaaagggccctcagaaaaaggagct agcaaaagaagaccaggtgccaatgagaagaataaactggagatgaataaagtcacagtaagata ttaaatactcctaacaatatcttctgttaatttatacagcatCATATGCTTCGGGCTCTC

FIG. 5TTT

[illegible]

FIG. 5UU

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
GH1u1	GH1	G	A	-	-	noncoding	GE527	AAGGGCCAGG GTATAAAA	TCTCCCTCAG GACACATTG	AAGGGCCAGGTTATAAAGggggcccaagagacc(g/a)gctcaaggatcccaaggcccaact ccccgaaccactcagggtcctctggagacagctcacctagctgcaatggctacaggttaagcgccct aaatccctttgggcaCAATGTGTCTGAGGGGAGA	166
GH1u2	GH1	A	C	-	-	noncoding	GE527	AAGGGCCAGG GTATAAAA	TCTCCCTCAG GACACATTG	AAGGGCCAGGTTATAAAGggggcccaagagacgggtcaaggatcccaaggcccaactcccc g(a/c)accactcagggtcctctggagacagctcacctagctgcaatggctacaggttaagcgccct aaatccctttgggcaCAATGTGTCTGAGGGGAGA	166
GH1u3	GH1	T	G	-	-	noncoding	GE527	AAGGGCCAGG GTATAAAA	TCTCCCTCAG GACACATTG	AAGGGCCAGGTTATAAAGggggcccaagagacgggtcaaggatcccaaggcccaactcccc gaaccactcagggtcctctggagacagctcacctagctgcaatggctacaggttaagcgccct aaatccctttgggcaCAATGTGTCTGAGGGGAGA	166
GH1u4	GH1	C	G	S	C	cds	GE609	GGGGAGACCT GTAGTCAG	GTCAAGTGGGC TCCAGATT	GGGGAGACCTGTAGTCAGAGccccgggagacagagcccaatgcccctccctccctgcaagaac ctagagctgctccgcatct(c/g)ccctgctctcactcagtcgctggagcccgctgagcttcc tcaggagtgctctccgcaacagcctggtgtagggccctctgacagcaaacgctctatgacctccta aaggacctagaggaaggcatcccaacgctgctgagggtgggggtggcgctagggtcccccAATCT TGAGGCCCCACTGAC	275
GH1u5	GH1	A	T	-	-	noncoding	GE527	AAGGGCCAGG GTATAAAA	TCTCCCTCAG GACACATTG	AAGGGCCAGGTTATAAAGggggcccaagagacgggtc(c/a)laggatcccaaggcccaact ccccgaaccactcagggtcctctggagacagctcacctagctgcaatggctacaggttaagcgccct aaatccctttgggcaCAATGTGTCTGAGGGGAGA	166
GH1u6	GH1	G	C	-	-	noncoding	GE527	AAGGGCCAGG GTATAAAA	TCTCCCTCAG GACACATTG	AAGGGCCAGGTTATAAAGggggcccaagagacgggtc(a/g)atcccaaggcccaact ccccgaaccactcagggtcctctggagacagctcacctagctgcaatggctacaggttaagcgccct aaatccctttgggcaCAATGTGTCTGAGGGGAGA	166
GH1u9	GHR	C	G	-	-	noncoding	GE602	GCTACAACATG ATTTTGGAAC A	GCTTCCCATTT TATTTAGTCT	GCTACAACATGATTTTGGAACAAatattctttttaaacccttcaatgtttaggacacactcaagaa tggactcaagaatggaaagaatgcctgattatgtttctctgctggggaacacagctgttactttaa tccatcgtttaccctccatctggataaccttatgtatcaagctaacacagcaatgggtggtacagtg atgaaaagtgttctctgttgatgaatatagglaaatcaacaggtttttgttttcatgtgacatagtt t(c/g)AGACTAAATAATGGGAAGC	287
GH1u7	GHR	G	A	R	H	cds	GE597	TTGAGTTGTTG ACTCTTGGC	TGACAAAGCC AGGTTAGC	TTGAGTTGTTGACTCTTGGCCaatatggcggtttatatatttctctgaaagatgggacccctatat tgacaacatacagttccagtgtagtactaatgaaagtggaatgaaggaatgaaggtgc(g/a)tgtag atccaaacaacgaacacictggaaattatggcgagttcagtgaggtgctctatgtaacacttccctc agatgagcccaatttacatgtgaaagggttaaaagaaataaaagattaaatagtaGCTAACCTGG CTTTGTCA	269
GH1u8	GHR	C	T	-	-	noncoding	GE597	TTGAGTTGTTG ACTCTTGGC	TGACAAAGCC AGGTTAGC	TTGAGTTGTTGACTCTTGGC(c/t)laatatggcggtttatatatttctctgaaagatgggacccct atatgacaacacatcagttccagtgtagtactaatgaaagtggaatgaaggaatgaaggtgcgtgtag atccaaacaacgaacacictggaaattatggcgagttcagtgaggtgctctatgtaacacttccctc agatgagcccaatttacatgtgaaagggttaaaagaaataaaagattaaatagtaGCTAACCTGG CTTTGTCA	269
GH1u1	GHR	A	G	G	G	cds	GE596	TTAAATTTGTT CTGCTGTGTA CT	GAAAGAAAGT CAAAGTGAAG G	TTAAATTTGTTCTGCTGTGTTACTTaattgctctgttgaattgacagtgcaaccagatccaccat tgccctcaactcaggacttactgaacgtcagtttaactgggatttcagcagatatccaaagtggat gggaagcaccacgcaatcagatatcagaaagg(a/g)tgaggtgtctggaggtatgaacttca atacaagaagtaaatgaactaaatggaaataatggaaataatggtaagatgttggctacaCCTTACACTTTGACT TTTCTTTC	268

FIG. 5VVV

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
GHRu2	GHR	A	C	I	L	cds	GE649	GCCCATATTCA GCTAAGCAAT	ATTGCCCCAGT CAATTCTTT	GCCCATATTTCAGCTAAGCAATCCCAAGTCCACTGTCACACATCGACTTTTATGCCAGGTGAGCGA 569 cattacaccagcaggtagtggtgctcttcccgggccaaagaaatgaaggaggtgtcccaat tgacatgaccccggaatgggtcactctccagagaaactctctatggacaatgctacttc gtgaggcagatgccaaaagtc[a/c]tccctgtgctctccacatcaaggttgaatcacaca tacgccaagcttaaaccaagagagacattacatccacagaaagccttaccactgctgctggg aggctgggacagagagacatgttccagggtctgagatgctgtccagactatccctccattca tatagtagctcccaacagggcctcactcaatgctgctgctgctgctgctgacaaagagt ttctctcatcatgtggtatgtgagcagacacacgaacaaatcatgcttagccttctctt gggtcccaagagctacgtatttataagcAAGAAATTGACTGGGCAAT
GHRu3	GHR	G	A	R	H	cds	GE596	TAAATTGTGT CTGTCTGTGTA CT	GAAAGAAAGT CAAAGTGTAAG G	TAAATTGTGTCTGTGTACTAATgctgtgtgattgacagtgcaacagatccacccat 268 tgccctcaactggactttactgaagctcagtttaactgggatttcagagatcccaagtggat gggaagcaccac[g/a]caatgcagatattcagaaagatggatggttcttgagttatgaacttca atacaagaagtaaatgaactaaatggaaatggtaagatgttgctacaCCTTACACTTGTGACT TTCTTTC
GHRu4	GHR	G	T	C	F	cds	GE1207	GCCATTTCATGA TAGCTATAAAC C	CTCACCTGGGC ATAAAGT	GCCATTTCATGATAGCTATAAACCCgaattccacagtgatgactcttgggttgaatttattgagct 498 agatatgtatgagccagatgaagagactgggaatccagacacagacagacttctaaagcagtgacc atgagaaatcacatagtaacctagggtgaggtgaggtgaggtcctggagctaccagctgtgtgaa cctgacattcttgagagactgttcaatgccatgacatcacatgaggtaccacagaggtgctca gccacagaggttaaaaggggaagcagatcttctgcttgacacagaaatcaaaataactcac cttatcatgtgctt[g/t]ccctgctactgagcagccagctgttatccaaagcagagaaacaa accacaaaccttccctactgaagagctgagtcactcaccaagctgcccataatcagctaaagca atccaaagtccactgtcaaacatcgACTTTTATGCCCGGTGAG
GHRu5	GHR	C	A	P	T	cds	GE649	GCCCATATTCA GCTAAGCAAT	ATTGCCCCAGT CAATTCTTT	GCCCATATTTCAGCTAAGCAATCCCAAGTCCACTGTCACACATCGACTTTTATGCCAGGTGAGCGA 569 cattaca[c/a]cagcaggtagtggtgctcttcccgggccaaagaaatgaaggaggtgtctcc caatgacatgaccccggaatgtgtcctctccagagaaactctctatggacaatgctca ctctgtgaggcagatgccaaaagtgcatccctgtgctctccacatcaaggttgaatcacaca tacagccaagcttaaaccaagagacattacatccacagaaagccttaccactgctgctggg aggctgggacagagagacatgttccagggtctgagatgctgctgctgctgctgacaaagagt tatagtagctcccaacagggcctcactcaatgagactgctgctgctgctgctgacaaagagt ttctctcatcatgtggtatgtgagcagacagacacacgaacaaatcatgcttagccttctctt gggtcccaagagctacgtatttataagcAAGAAATTGACTGGGCAAT
GHRu6	GHR	C	A	P	T	cds	GE649	GCCCATATTCA GCTAAGCAAT	ATTGCCCCAGT CAATTCTTT	GCCCATATTTCAGCTAAGCAATCCCAAGTCCACTGTCACACATCGACTTTTATGCCAGGTGAGCGA 569 cattacaccagcaggtagtggtgctcttcccgggccaaagaaatgaaggaggtgtcccaat tgacatgaccccggaatgggtcactctccagagaaactctctatggacaatgctacttc gtgaggcagatgccaaaagtgcatccctgtgctctccacatcaaggttgaatcacacataca gccaaagcttaaaccaagagagacattacatccacagaaagccttaccactgctgctggagg c/a]ctgggacagagagacatgttccagggtctgagatgctgtccagactatccctccattca tatagtagctcccaacagggcctcactcaatgagctgctgctgctgctgctgacaaagagt ttctctcatcatgtggtatgtgagcagacacacacgaacaaatcatgcttagccttctctt gggtcccaagagctacgtatttataagcAAGAAATTGACTGGGCAAT
GHRu1	GHR	G	C	W	S	cds	GE599	TCAACCTTGTG TGGATCTAATT T	GGGGCTATCCT GAATGTTAAT A	TCAACCTTGTGCTGGATTAATTgattgtgatttcattgcttagaatgaagcacaattcaaaa 269 ctctagctggccttattctactgactt[g/c]gtgctggaagctgctccagcagactgggt cctatggactgcgcccggaggaagagagatgccgaatattgattgattcttccaaagggt agttctctcagcttcaaaataagacatagtagtttggattcaatttaactatATTAAACATTCAG GATAGCCCC

FIG. 5WWW

81/178

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
GNRHRu 2	GNRHR	C	T	F	F	cds	GE569	GGAAACACCA TTTCATTTCT	CAAGGCTTAT CCACCTCTA	GGAAACACCATTTTCATTTCTTTTctccatctcaagagcatcacatctctcttcttcagatagtc aaagaggttggtcaactggcagaaacccacgctt [c/t] gaatgcaccacgcaccagccacgctt ctccctccgagagactgaagagagctctggttaagttaagtgcatacaatgatcacacagcatag agctctAGAGGTGGATAAGCCTTTG
GNRHRu 3	GNRHR	A	C	-	-	noncoding	GE578	CATTAAAGGG TTTATGTGAGG AT	TCTAAAGAAGA AAACTCGTGC TA	CATTAAAGGGCTTTATGTGAGGATTTTaaataattacattaaaaaaagagcatagtcatt tgcagataaattaccagcaggaagaatttcaatgtctctggaataattccctataaaaaagaga taggaacacagaaagtcacagtaacacactcaaggaagattgggatcttttggctct ctgctctaaacaggtaa [a/c] aggtcttgattattctTAGCACGAGTTTCTTTTAGA
GNRHRu 4	GNRHR	C	A	S	Y	cds	GE599	TCAACCTTGTC TGGATCTAATT T	GGGGCTATCCT GAATGTTTAAT A	TCAACCTTGTCGTGATCTAATTgatttgctcattctgctttagaatgaagcccaattcaaaaa ctctagctggccttattctactgacttggtgcgtggaaggtctgctcagccagcactggtccta tggactgcgctccctgggaagagagatgccgaataattgattctctccaaag [g/t] ta agttctctcagcttcaaaaataagacatagtgatttgattcaatttaactaTATTAAACATTCAG GATAGCCCC
GNRHRu 5	GNRHR	G	T	-	-	noncoding	GE599	TCAACCTTGTC TGGATCTAATT T	GGGGCTATCCT GAATGTTTAAT A	TCAACCTTGTCGTGATCTAATTgatttgctcattctgctttagaatgaagcccaattcaaaaa ctctagctggccttattctactgacttggtgcgtggaaggtctgctcagccagcactggtccta tggactgcgctccctgggaagagagatgccgaataattgattctctccaaag [g/t] ta agttctctcagcttcaaaaataagacatagtgatttgattcaatttaactaTATTAAACATTCAG GATAGCCCC
GPIBA 2	GPIBA	C	T	T	M	cds	GE493	CTTCCAGGGGA TGCAGG	AAAGCAAAAG GCAGGAGGT	CTTCCAGGGGATGACGGGGatccactcaaggctcccttgccacaggtcctcatgctctctc ctcttgctgctctcctgctgcaagcccttaccacccaccccatctgtgaggtctccaaagtggc cagccacctagaagtgaaactgtgacagaggaatctgacagcgtcctccagacctgccgaag acacacccatctctccactgagtgagaaactctctgacaccttctccctggcaacctgtgctt taactcgcctcactcagctgaacctagatagtgagagctcaccagctccaggtcagtgaggac gctgcagtgctgggaacctggatctatccacacatcagctgcaagcctgctgctgctgagggc agacactgctgctcaccgtcctggacgtctcctcaacccgctgcaagcctgctgctgctgctgct gctgcgtggtcttgccgaactccagagctctaccctgaagagctcagctgagctgaagacccctg cccagggctcctga [c/t] gcccacacccacagctgagagagctcagctggtctaaacaaacttg actgagctcccgctgggtcctggaatgggtgagagatctcgacacccctctcctccaaagaa ctcgtgtatatacaataacaaagggttttttgggtcccACCTCTCTGCTTTTGTCTTT
GPIBA 3	GPIBA	C	T	N	N	cds	GE495	GACACCTTCT CCTCCAAGA	GATTGGGTGG GCTCCG	GACACCTTCTCTCAAGAGaaactcgtgtatatacaataacaaagggttttttgggtcccacct cctgcttttctgtcttctccacgggaacccctggttatgcaactgtgagatcctctatttctgctc gctgctgcaggaacaatgctgaataatgtctacgtatggaagcaaggtgtggacgtcaaggccatg acctctaa [c/t] gggccagtgctgagtgacaaatcagacaagttcccgctcacaataacc caggaaggggtgccccacccttggtgagtgaggtgacacagacctatagttactaccagaa gaggacactgaggggataaggtgcgtgccacaagagctggtcaagttccccacaaagccca taacacccctgggtctattctactcattggtccactgctctctagacagccaaatgctctcct ccttgcatccaacacagaatccactaaggagcagacacattcccactagatggaccccaaat ttcacacttcacatggaatccatcacattctccaaaactccaaatccactactgaacccacccc aagcccgaccacctcagagccgtcccgagggccgcccccaacatgacacccctggagccactc caagcccgaccacccccagagccccactcagagccccccccccagccccccCGGAGCCCCACC CCATC

FIG. 5XXX

[illegible]

FIG. 5YYY

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
GP9u4	GP9	G	A	A	T	cds	GE401	CTACATCCCA GTGCTGC	TCTGTGGTTT GGGCTGAC	CTACATCCCAAGTGTGCTGCgttccctgaggatcgggtccagggtgcagggccctccctccacagcccctctctctgcagccagcctgtccatgctcctggtggagccctgttctgtctgtgggcaacagc agggccacaaggactgccccagcccatgtacctgcccgtggaacccatgggctgtgggtgaactgcaggggccacggactacg [g/a] cccctgctgcccgcacccacaccccttctgtgtggcacaacacagccttcagtcggtccgccccggagcccttgaccactatgtgagccttctgtgg agacctgattgacgagaaacccctggactgtgactgcagcctcaactatctgccccctgtgg ctggaggaccgcacgcccaggccctgctgeaggtccgctgtgccagccccagctcgtgtgccat tggcccgtctggccggctgtgacagggtaccaggctggcgtggcagctggcagcggtccct ggggtgccccgggggtctctgtggagcgtggcgctgtgtcccggtggccgctgggctgtctct ctggctggcctgtgtgtgccccacagggccctggatgagccagggcccccaagacctggctc cagccaggggggcagtcctcctgagggtccccagactccaccaagctgtgtcagcccaaacac CAGA	719
GP9u5	GP9	G	A	A	T	cds	GE401	CTACATCCCA GTGCTGC	TCTGTGGTTT GGGCTGAC	CTACATCCCAAGTGTGCTGCgttccctgaggatcgggtccagggtgcagggccctccctccacagcccctctctctgcagccagcctgtccatgctcctggtggagccctgttctgtctgtgggcaacagc agggccacaaggactgccccagcccatgtacctgcccgtggaacccatgggctgtgggtgaactgcaggggccacggactacg [g/a] cccctgctgcccgcacccacaccccttctgtgtggcacaacacagccttcagtcggtccgccccggagcccttgaccactatgtgagccttctgtgg agacctgattgacgagaaacccctggactgtgactgcagcctcaactatctgccccctgtgg ctggaggaccgcacgcccaggccctgctgeaggtccgctgtgccagccccagctcgtgtgccat tggcccgtctggccggctgtgacagggtaccaggctggcgtggcagctggcagcggtccct ggggtgccccgggggtctctgtggagcgtggcgctgtgtcccggtggccgctgggctgtctct ctggctggcctgtgtgtgccccacagggccctggatgagccagggcccccaagacctggctc cagccaggggggcagtcctcctgagggtccccagactccaccaagctgtgtcagcccaaacac CAGA	719
GRF1	GRF	C	T	L	F	cds	GE510	AACACACAGA GAGAGCAAC	ACACCCATACC TGTGCTT	AACACACAGAGAGAGCAACaaagcagaggaggaagggcaagggcaag [c/t] ttggctgtcaggtaga cagcatgttgggcagaaacaaagcaaatggaaattggagagagatcctgttggccctgtgcagaaagc ACAGGTATGGGTGT	144
GRIN1u1	GRIN1	T	A	I	N	cds	GE1121	AGTCTGGGCC TTGGCG	GAGTGACCCCG CCCACC	AGTGTGGGCCCTTGGCGgggtccccgaacggggagggaacccccacgggtctgtagtgcctgcgc ctaggga [t/a] cctcgggtgcagctcatcaacggcaagagagtcggcccaactcagcgacgc cgttggcgctgtgtggcccgccgtgcacagctcctcggagaaggagacatcacgcagccgcgc cgggtgcgttgggcaacacacacatctggaaagccggcgctctcaayaggtggggcggggcc tcccgaggcttggggcgggggtgtctcttggggaggTGGCGGGGTGCTC	309
GRIN1u2	GRIN1	G	C	D	H	cds	GE1293	CCCCCGCAGAC AGACAG	CCTTCCCGAGC AGCAGG	CCCCCGCAGACAGACAGacagcggattgggacagcgcccgccccacgcagagagccccggagcacc acggggctcggggagagagacccccccctccccaggtcgcgcctggcccgcccggttggcc cgtggcccggtccaccccgctcccggcccccgctgtgccccagcgttggggctaaacggggcgccctt gctgtgtattctctattttgagcaggtaccatcccatgatcacgggcccgcgtcaacctctca [g/c] atccccctcagcaacccgtgtgtgagggcccccgggcgccccacctgtgccaggttagccccg gccaaggacactgtgggtCTGCTGCTCGGGAAGG	361
GRIN1u3	GRIN1	A	G	P	P	cds	GE1085	GAGGACCTGGG CCTGCG	AGCCCGTGGG TCCTCC	GAGGACCTGGGCTGGGagcgcgggttggagtgctggagtcctggccgctcatccccgctcgc cccacagcagagacgattgctgccactgtataccgcgcagccgcgattgtaacatgacgggtc cggtaacgtgtgtgtgtgtgtggcgagcgagatctcggggaaacgcgcctgtgcgtacgcccc [a/g] gacgctgagtgctgtggggttggcggggtccccgaacggggagcccccaacgggct	250

FIG. 5AAAA

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
GRIN1u4	GRIN1	G	C	E	Q	cds	GE1287	GAGCTGAGAG AGACTGCCG	GTAGGTGCGG CGGTAAC	GAGCTGAGAGAGAGACTGCCGccctgggagccttaggtcgggtggtccaggctgggtctcccttc ccccccagattgtgagcgtccacag(g/c)agccctcgtgtcgtcagtcagccacagcgtgagtga tggacatgcaaggagaggttccaggtcaacggcgacccagtcaggaaggtgatctgcacgggc cacaagacagtcgcccgggagcagcgtgagtcgcccgggagcagggcgccggcgccggcgccg ggggcgtggggcgtctggagcccgagcagttACCGCCCGCACCTAC	306
GRIN1u5	GRIN1	T	G	C	G	cds	GE1287	GAGCTGAGAGAG AGACTGCCG	GTAGGTGCGG CGGTAAC	GAGCTGAGAGAGAGACTGCCGccctgggagccttaggtcgggtggtccaggctgggtctcccttc ccccccagattgtgagcgtccacagcagcgtcgtgtacgtcagccacagcgtgagtggg aca(t/g)gcaaggagaggttccaggtcaacggcgacccagtcaggaaggtgatctgcacgggc ccacgacacgtcgcggcgagcagcgtgagtcgcccgggagcagggcgccggcgccggcgccg ggggcgtggggcgtctggagcccgagcagttACCGCCCGCACCTAC	306
GRIN1u6	GRIN1	G	C	D	H	cds	GE1287	GAGCTGAGAGAG AGACTGCCG	GTAGGTGCGG CGGTAAC	GAGCTGAGAGAGAGACTGCCGccctgggagccttaggtcgggtggtccaggctgggtctcccttc ccccccagattgtgagcgtccacagcagcgtcgtgtacgtcagccacagcgtgagtggg acatgcaaggagaggttccaggtcaacggcgacccagtcaggaaggtgatctgcacgggc c(g/c)acagtcgcccgggagcagcgtgagtcgcccgggagcagggcgccggcgccggcgccg ggggcgtggggcgtctggagcccgagcagttACCGCCCGCACCTAC	306
GRIN1u7	GRIN1	G	A	K	K	cds	GE115	GCGGAGCTGG GAGGAC	AGGGACGGAG GTCAGC	GCGGAGCTGGGAGGAGCgctgctgcatgcccggcgtctgtcgcctcgcaggtgaaacacagc aacaagagaggtggaatgggagtgatggcgagcgtcgcagcggcgagcagcagcagcagcagc gcccataacacacagcagcgcgcagtcacagtcaggttttccaaagccctcagtcacagc gctgactattctggtcaa(g/a)laaggtggcgagggcgccgggtggcggggtggcgggggga gtccctggagggcccgccgctgacccctgacccctgacccctgacccctgacccctgacccctg	299
GRIN1u8	GRIN1	G	A	E	E	cds	GE1120	TTCCGCGAGTG GGAGGC	CGTCTGACCC TCGGCT	TTCCGCGAGTGCGGAGGCGgggtgggagggcggtccccgggggtccacccctcagccacccgctgccc cgctcccgagagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagcagc cgctgggttgagatgagtcggtgttcacagctgggaacacacacacacacacacacacacacac acga(g/a)ggcgggcgctcagaaacgcctggagacgcctgctgagggagcgtgagtcacaggt gaggtcggcgccgctggggcgtggcgccctggcgagcagcagcagcagcagcagcagcagcagc	309
GRIN1u7	GRL	G	T	-	-	noncoding	GE1196	ATTCAGGTGG CCAAATTAT	AAAGAAACAA AAACATGTC	ATTCAGGTGGCCAAATTATtttggttaataagaaactgaaatactataataataataataata tcaataataattttatatttagttatagttttagatataataataataataataataataataata gaaggaggggctactgagcgtttacatgcaattttataataatgattgtaataatagcttggatag tgtaataataagaaatgatttttagatgagattgtttatcatgacatgattataattttttgagg (g/l)gtcaagaaatgctgagttggtccttagggagagagagagagagagagagagagagagag gcagcaggtgctcagaaacacacagtttggcttagggagagagagagagagagagagagagagag tgtagagtgagggttctgagcgtcgtgacccagtgagattacagcagagagagagagagagagag cattctgacaccccttctcattccacagtgagtcgtcagcagcaggttagttactcaatctc ccctgcaactaaagtagtaagtagtgaacagagagagagagagagagagagagagagagagag ggcaccatcaatagcgggttacttccacacagcagcagcagcagcagcagcagcagcagcagcag gcttcagaaagttggcaatagttgcatagaggtaccagcaataataataataataataataata taggtgccaataataacataattctcttctctacacacacagagagagagagagagagagagag agggacatggtttgtttgttttttt	802
GRL18	GRL	G	T	-	-	noncoding	GE628	GACCTTCCCAT TACAGTATTC	GAAGAAACAC AAAGGTTTATA TAGTTGC	GACCTTCCCATTACAGTATTCtatgtatt(g/t)tttaataccacacagctcgaatacaaa agaaataataaaggaattccagcgggccaacacagagagagagagagagagagagagagagag ggtaacaaacaaatagttccctgcaaggttaccacacacacacacacacacacacacacacac ggttatgaaactgaaggttatagcag tcagactacgctcaacatggttagggggcgagagagagagagagagagagagagagagagagag ataccaggtgaagatgcaaacacataaagagAACATATATATAACCTTTGTGTTTTC	383

FIG. 5B BBB

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
GRRLu11	GRL	A	C	-	-	noncoding	GE558	AAATATGTTTG AAGACCTGTGA A	AAATGATAAAT TTTTATAAGCT ACAGAG	AAATATGTTTGAAGACCTGTGAACCTTCAATAGTGCCTTTTATCCATAT [a/c]ggacagcaca attacctatgtctgagggaatgatcgcatcatcgatacaaaatcgagaagaaaaaactgcccagca tgcgctatcgaaaaatgtcttcaggctggaatgaacctggaaggtaatataaataatctgaaagca attgttctctgtagcttatataaaatttatcatt
GRRLu12	GRL	C	T	H	H	cds	GE565	CTTCTGAAGAG TGTTGCCCTCAT	GGGAAAATGAC ACACATACAAC T	CTTCTGAAGAGTGTGCCCTCATACCTTTATTTCTCTTAATTCAGGTTTCAGGAACATTACA [c/t] ctggatgaccaaataagacctactgactcagactcctggatgttcttatggcatctgctctgggtg gagatcatagacaatacaagtgcacaacctgctgtgttttgcctcctgatctgatttataatgagt aAGTTGATGTGTGTCATTTC
GRRLu13	GRL	T	A	-	-	noncoding	GE1325	TGATCTGTCAA ACTTCCAGAAC C	TTGGCATGTCT GTAAATG	TGATCTGTCAAACCTTCCAGAACCATggttagccttcagtgagatttccatcttggctggctcactcc ctgactgtagctgtagggaatgtgttttctgtgtgtctgtgttctgtgttcagtgatcagaagggaa ataaagtgtgaaggagacactt [t/a]aaaccttgggtggagtttcgttaatttcccagacta tttcaagcaacctggtccaccagattagtgaccaggttttcaggaaaagatttgcctctctc tagaaaatctgtgaaagattttatttctgtgtagaaggtgtgtaaaataacctcctcaata acttgcttaactacatagattcaagtgtgtcaattcttatttctgtatataatgctatata atggggacaatctatatatactgtgtgtgacattataagaagcttttctcatattttttatc acagtaattttaaaatgtgtlaaaaattaaaaccagtgactcctgttttaaaaaataaaagtgtgagt tttttatctgctgaataataatctgttagttaaaaaaaggtgtcttttacctacgcagtgaa atgtcagactgtaaaacctgtgtggaatgtttaacctttatttttcttcaaatgtgtgt ctggtattaccaaaaccacacattgtaccgaattggcagtaaatgttagcattTACAGCAATGC CAA
GRRLu14	GRL	G	C	D	H	cds	GE1201	CAACGGTGGCA ATGTGAAA	AAGAAACAGGA AAACACTGAT	CAACGGTGCAATGTGAATgtataccacagaccacacacctttgacattttgcaggatttgg agttttctcttctgggtcccccaggtaaaggacgaatgagagtccttggagatcagacctgtgata gatgaaaactgttgtcttctctctctgtggggagagacagattcttcttttggaggaaactc gaatgaggactgcagacctctcatcttaccggacactaaaccacaaataaggaataatggagatc tggttttgtcaagccccagtaattgtaacactgcccccaagtgaacacagaaaaaagaagatttcatc gaactctgcacccctgggttaattaaagcagagaaaactgggcacagtttactgtcaggcgaagctt tcttgagcaaatataattggttaataaaatgtctgccatttctgttcaatgtgtgagtaacctctg gaggacagatgaccactatgacatgaatacacagatccctttctcaacagcag [g/c]atcagaa gcctattttaaagtcatccacaaatccctgttgcgaataatgggaatagggtgccaaaggt ctggagatgacaacttgacttctctgggactctgaacttccctgggtcgaacagttttttctaat ggctattcaagggttaagATCAGTGTTTTCTGTTCTT
GRRLu15	GRL	C	A	-	-	noncoding	GE664	GCATTTTGTGAT TTATGCATGG	GAGGAATTACT TTGTCTGATTA AAA	GCATTTTGTGATTTATGCATGGAaacctgaaaaaaagtttacaagtgatatcagaaaaagggaggt tgtgccttttatagctattactgtctgtgttttaacaaattcttcttataatttagtgaactacgctt gtcattttttcttacaataattttttatcaagttattgtacagctgttttaagatgggcagctag ttcgtagctttcccaataaaactcaaacattaaatcaatctctgtgtgaaatgggttgggtgtct tcaaaccttgatggcacttagctatcagaagaccacaaaaattgactcaaatctccagtaattctt9 tcaaaaaaaataaaagctcatatttgtatatatactgtctcagtgagaaattatattaggt tgtgcaaatcaacagtcctaa [c/a]tggtatagatgactgtccagtgacctgctgggtaaac tgtggatgattgttgcaaaagactaatttaaaaaataaataccaagagccctgtctgtacctaa cgccctattttgtcaatggctatatggcagaagagctgtgtaaaactattgtcttccaggacctt tgaagtagtttgtataacttcttaaaagtgtgtatccagataaccagctgttaacacagctgaga gactTTTAAATCAGACAAAGTAATTCCTC

FIG. 5DDDD

[illegible]

[illegible]

FIG. 5FFF

FIG. 5GGGG

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
HSD3B1 d86	HSD3B 1	T	C	F	L	cds	GE675	GGTGGCACCTCTTAGGGGATA	GGCGGGTGCGATAGGTGTAAA	GGTTGGCACCTCTTAGGGGATAatctctgacagtgacaatattgctcttctcatggtgacagtgacaggtaccaggtatctctggttagagggctgtgtccaaagctagtgccagtgctcatctacacagtagcatagaggtatgcccggcccaactctacaaggaaatcatccagaatggccatgaagaagagccctctggaacaacatggcccgctccatacccacacagcaaaagcttgcctgagaaggtctgactggtgaggtacaggggtggaattctgaaacacggcgccacccctgtacactgtgacctgacgacccatgtatctatctatgaggaaggaagccgattctctgtagtataaacgagggccctgacacaacatgggatcctgtcaagtgttgaaagtctctccactgttaacccagtcatagttggaatgtggccctgggcccacattctggcccttgaggccctgcaggaccccaaggagggcccaagctccggagacagttctactatctacagatgacacgctcaccaaaagctatgataaccttaattacacccctgagcaaaaggttcggccctccgcttgattccagatggagtc/c/ttcccttctccctgattgattggattggcttctctgctggaatagtgagcttctactacaggccaatttTACACCTATCGACCGGCC	692
HSD3B2 d25	HSD3B 2	G	A	A	A	cds	GE1194	CAGAAGATGCAACCTGAGTCA	GCCAGATCTCGCTGAGCC	CAGAAGATGCAACCTGAGTCTataacaaccacaccacggaggaggaggagggggacacaagcagcagcagggctggcagcacctccgggagaaatattctcacacaacacatcatctctctgtggcaggtaccagctactgttggaggcctgtgtccaaagccagtggtccagtgcttcatctacacagtagcagtagaggttagcggcccgcccaactctcacaggaaatcatctcagaaagcccaagaaagagcctcttggaacacatggcccaactccataccgtacagcaaaaagcttgctgagaaaggtgtgctggc[tg/a]gctaatgggtggaattctaaaaatgggtatcccttgtaacctgtgcttgaagccccacatatattctatggggaaggagggcccatctcttctgcccagataaaatgaggccctgaacaacaatgggatccgtcaagtgttggaagtctctccacagtcacccacagtcagctatgttggaacgtggcctgggcccacattctggccttgaggccctgcaggagcccaagaggcccaagcatccgaggagcaaatctctacatctcagatcacgcctcccaaaagctatgataaccttaattacacccctgagcaaaaggttcggccctccgcttgattccagatggagcctctctttaaaccttgatgtactggattggcttctctgcctggaaatagtgagcttctactacgcccacatttacacctataaccccccttcaaccgcccacacagtcacattatcaaatagcgtattcaccttctcttacaagaaGGCTCAGCGAGATCTGGC	839
HSD3B2 d26	HSD3B 2	A	G	T	T	cds	GE1194	CAGAAGATGCAACCTGAGTCA	GCCAGATCTCGCTGAGCC	CAGAAGATGCAACCTGAGTCTataacaaccacaccacggaggaggaggagggggacacaagcagcagcagggctggcagcacctccgggagaaatattctcacacaacacatcatctctctgtggcaggtaccagctactgttggaggcctgtgtccaaagccagtggtccagtgcttcatctacacagtagcagtagaggttagcggcccgcccaactctctacaaggaaatcatctcagaaagcccaagaaagagcctcttggaacacatggcccaactccatagcgtacagcaaaaagcttgctgagaaaggtgtgctggggccttaattgggtggaattcaaaaaatgggtataccttgtaaccttgtaagccccac(a/g)tatattctatggggaaggagggcccatctcttctgcccagataaaatgaggccctgaacaacaatgggatcctgtcaagtgttgaaagttctccacagtcacccagctatgttggaacgtggcctgggcccacattctggccttgaggccctgcaggagcccaagaggcccaagcatccgaggagcaaatctctacatctcagatgacgcctcccaaaagctatgataaccttaattacacccctgagcaaaaggttcggccctccgcttgattccagatggagcctctctttaaaccttgatgtactggattggcttctctgcctggaaatagtgagcttctactacgcccacatttacacctataaccccccttcaaccgcccacacagtcacattatcaaatagcgtattcaccttctcttacaagaaGGCTCAGCGAGATCTGGC	839
HSD3B2 d27	HSD3B 2	C	T	-	-	noncoding	GE639	AAAAATAGGCACTCTGCTGAGTGAT	CCATGCAGACTTTAAGATGGAGTAT	AAAAATAGGCACTCTGCTGAGTGATaaccattttacctctgttttttagccctctctctgggtcacgctagaatcagatctgctctccagatcttctgttctctggg[c/t]aagtgcttctctgctacttttggttggccacgatgacgggctggagctgacctgtgtgacaggggcagggagggctctctgggtccagagatcgctccgctgttgggtggaagaggaagaaactgaactgaaggagccttggacaaggccctcagaccagaatttgagaggaatttctctagtaagtaaaacttgagtcattgggtctctgtggcttcttTAACTCTGCAATGG	338

FIG. 51111

SUBSTITUTE SHEET (RULE 26)

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' → 3')	Reverse Primer (5' → 3')	Assay Sequence
HSD3B2 u15	HSD3B 2	C	T	-	-	noncoding	GE665	CCTGCTGGAAA TAGTGAGCTTC	GCTCTTTTGT TGAAGTGTGTG AA	CCTGCTGGAAATAGTGAGCTTCctactagcccaatttacacctatcaaccccccttcaaccgccc acacagtcacattatcaaatagcgtatcaccttctctacaagaaggtcagcgagatctggcg tataagccactctacagctggagggaagcagcaaaacccgtgggtgggttcccttgg ggaccggcacagagagaccctgaagtcgaagactcagtgatttaaggatgacagagatgtgcatg tgggtattgttaggaaatgtcatcaactccaccctggcctcacagaagcaacaaagggc acaagcccaggctcctgctgctcctttcacacaatgcccaacttactgtattcctcatgtcatc aaaa(c/t)ctgcacagtcactggcccaacaagacgttctctcctaatacacacagaagaa aaacaatatgatttgcgtttaccaaattctcagtagctgattctgaacaatttgaggacccttaa actgaaggggcccttttgactaatagagctccatttccactcttaaatgagaagcatttcccttc tctttaatctcccatctctCACACAGTTCACAAAAGAGC
HSD3B2 u16	HSD3B 2	A	C	-	-	noncoding	GE665	CCTGCTGGAAA TAGTGAGCTTC	GCTCTTTTGT TGAAGTGTGTG AA	CCTGCTGGAAATAGTGAGCTTCctactagcccaatttacacctatcaaccccccttcaaccgccc acacagtcacattatcaaatagcgtatcaccttctctacaagaaggtcagcgagatctggcg tataagccactctacagctggagggaagcagcaaaacccgtgggtgggttcccttgg ggaccggcacagagagaccctgaagtcgaagactcagtgatttaaggatgacagagatgtgcatg tgggtattgttaggaaatgtcatcaactccaccctggcctcacagaagcaacaaagggc acaagcccaggctcctgctgctcctttcacacaatgcccaacttactgtattcctcatgtcatc aaaa(c/t)ctgcacagtcactggcccaacaagacgttctctcctaatacacacagaagaa aaacaatatgatttgcgtttaccaaattctcagtagctgattctgaacaatttgaggacccttaa actgaaggggcccttttgactaatagagctccatttccactcttaaatgagaagcatttcccttc tctttaatctcccatctctCACACAGTTCACAAAAGAGC
HSD3B2 u17	HSD3B 2	G	T	L	L	cds	GE1194	CAGAAGAATGC ACCCCTGAGTC	GCCGATCTCG CTGAGCC	CAGAAGAATGCACCCCTGAGTCTataacaacacacacacagggaggaggggacacaagca ggcagcagtggtggcagcactccgggaggaatacctcacacacacacacacacacacacagga taccagactact(g/t)ttggaggctgtgtccaaagccagtggtgagcttctcatcacacaggt agcatagaggtagcggcccaactcctacaggaatacctcagcaagccacacaggaagagggc tcggaaac cggtaatgggtgggaatctaaataatgggtgataccttgcacacttgggtgaggaagctgtgctgg atctatggggaaggaggcccttctctgcccagtaataatgagggccctgacacacacacacacac cctgtcaagtgtggaaagtctccacagtcacacacacacacacacacacacacacacacacac acattctggccttgaggccctgcaggacacacacacacacacacacacacacacacacacacac taacatctcagatgac cggtcctccgcttgcattccagatggagcccttctttaaaccctgattgactggttggcttctgc tggaaatagtgagcttccactagcccaatttacacacacacacacacacacacacacacacacac gtcacattatcaaatagcgtatctacacttctcttacaagaagcctcagcagatctggtg
HSD3B2 u18	HSD3B 2	C	T	L	L	cds	GE665	CCTGCTGGAAA TAGTGAGCTTC	GCTCTTTTGT TGAAGTGTGTG AA	CCTGCTGGAAATAGTGAGCTTCctactagcccaatttacacctatcaaccccccttcaaccgccc acacagtcacattatcaaatagcgtatcaccttctctacaagaaggtcagcgagatctgtt ggcgtataagccactctacagctggagggaagcagcaaaacccgtgggtgggttcccttccc ttgtggaccggcacagagagaccctgaagtcgaagactcagtgatttaaggatgacagagatgtg catgtgggtattgttaggaaatgtcatcaactccaccctggcctcacagaagcaacaa ggcacaagcccaggctcctgctgctcctttcacacaatggcccaacttactgtattcctcatgt catcaaaaacccctgacagtcactggcccaacaagacgttctctcctaatacacacagaagaa aaacaatatgatttgcgtttaccaaattctcagtagctgattctgaacaatttgaggacccttaa actgaaggggcccttttgactaatagagctccatttccactcttaaatgagaagcatttcccttc tctttaatctcccatctctCACACAGTTCACAAAAGAGC

FIG. 5KKKK

[illegible]

FIG. 5MMMM

[illegible]

FIG. 5NNNN

SUBSTITUTE SHEET (RULE 26)

[illegible]

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
HTR1A u1	HTR1A	C	A	-	-	noncoding	GE1181	GACTGTTTGGCTAGTGGGGAGA	TGAGGAAGCCAATAAGCCAA	GACTGTTTGGCTAGTGGGGAGACTCCAGCTCCGCGCAGCCAGTTCGGGAGCGGCAAAAGTAAAATGGA	873
HTR1B u1	HTR1B	G	C	V	V	cds	GE1318	GGCCTTCTTCTGGCGTC	CCTACCTGTGGAACCAAGACA	GGCCTTCTTCTGGCGTCAGGCTAAGGCTGAGGAGGAGGAGTGTCTCGGAATGCGTGGTGAACACCGACCCACATCCTCTACACGTTACTCCACGGTGGTGGTCTTCTACTTCCACACCTGCTCCTCATCGCCCTCTATGGCGCATCTAGTAGAGCCGCTCCCGGATTTGAAACAGACGCGCAACACAGGACCGGCAAGCGCTGACCGGAGCCAGCTGATAACCGACTCCCGGGTCCACGTCCTCGGTCCACCTCTATAACTCGGGTCCCGACGTCGCGCAGCAATCCGGAATCCTCTCTGTGTG/c]AACCAAGTCAAAGTGGAGTCTCCGACGCTGCTGGAGAAAGAAAGAACTCATGGCCGCTAGGAGCGCAAAAGCCCAAAGACCTAGGATCATCTGCAAGATGCTGTGGTTCGCTACCTCTCTCATCATCAGCTAGTGATCTCACTCCCTCATCAACCCCATATCTATACCATGTCCAAATGAGGACTTAAATGAGCATCCATAAAGTATAGTGTGACAAAGTGTGACTGCGCTTTCGAGTGGGGTCCGCTAAGCGGACCTTGGGGACCAAGTGTGCTGCTTCCACAGGTAGG	698
HTR1B u2	HTR1B	G	A	-	-	noncoding	GE1318	GGCCTTCTTCTGGCGTC	CCTACCTGTGGAACCAAGACA	GGCCTTCTTCTGGCGTCAGGCTAAGGCTGAGGAGGAGGAGTGTCTCGGAATGCGTGGTGAACACCGACCCACATCCTCTACACGTTACTCCACGGTGGTGGTCTTCTACTTCCACACCTGCTCCTCATCGCCCTCTATGGCGCATCTAGTAGAGCCGCTCCCGGATTTGAAACAGACGCGCAACACAGGACCGGCAAGCGCTGACCGGAGCCAGCTGATAACCGACTCCCGGGTCCACGTCCTCGGTCCACCTCTATAACTCGGGTCCCGACGTCGCGCAGCAATCCGGAATCCTCTCTGTGTG/c]AACCAAGTCAAAGTGGAGTCTCCGACGCTGCTGGAGAAAGAAAGAACTCATGGCCGCTAGGAGCGCAAAAGCCCAAAGACCTAGGATCATCTGCAAGATGCTGTGGTTCGCTACCTCTCTCATCATCAGCTAGTGATCTCACTCCCTCATCAACCCCATATCTATACCATGTCCAAATGAGGACTTAAATGAGCATCCATAAAGTATAGTGTGACAAAGTGTGACTGCGCTTTCGAGTGGGGTCCGCTAAGCGGACCTTGGGGACCAAGTGTGCTGCTTCCACAGGTAGG	698

FIG. 5RRRR

FIG. 5SSS

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
HTR1Du 5	HTR1D	A	G	T	T	cds	GE1162	TTGAAGGAAGG AGCCAAATG	TGTAGGAGATC TGAGAGGTGTT CA	TTGAAGGAAGGAGCCAAATGTgtggagggtctgtggaagagagagagccacccatgcatgtccccact gaaccagtcagcagagaggccctcccccaggagccctccacacagatccctgaatgccacagaaacct cagaggcttgggataccaggaacctccaggcgctcaagatctcccttgcgtgggtcccttccgtc atcac(a/g)ctggccacagtcctctccaatgcttctactcaccacacatctactcaccagga agctccacacccctgccaaactaactgattggctccctggccacccacccaccccttggtttccatc ttggtaattggccatcagcatcgctataccatcaccacacacccctggaaactttggccaaatcttgg tgacatctgggtgtctcttgacatcacgtgctgacagccctccatctctctgtgtcatctg ctctggacaggtactgggcaatcacagatgcccctggaatacagtaaacgcagagcggtggccac gcggccacacatgctggccatgtctgggcatctccatctgcatctccatccccccgctctctg gcggcaggccaaaggccaggagagatgtcggaactgtctggTGAACACCTCTCAGATCTCTCTACA
HTR1Du 6	HTR1D	T	C	I	I	cds	GE1162	TTGAAGGAAGG AGCCAAATG	TGTAGGAGATC TGAGAGGTGTT CA	TTGAAGGAAGGAGCCAAATGTgtggagggtctgtggaagagagagccacccatgcatgtccccact gaaccagtcagcagagaggccctcccccaggagccctccacacagatccctgaatgccacagaaacct cagaggcttgggataccaggaacctccaggcgctcaagatctcccttgcgtgggtcccttccgtc atcacactggccacagtcctctccaatgcttctactcaccacacatcttactcaccaggaagct ccacacccctgccaaactaactgattgtctggctccctggccacccaccccttggtttccatc ttggtaattggccatcagcatcgctataccatcaccacacacccctggaaactttggccaaatcttgg tgacatctgggtgtctcttgacatcacgtgctgacagccctccatctctctgtgtcatctg ctctggacaggtactgggcaatcacagatgcccctggaatacagtaaacgcagagcggtggccac gcggccacacatgctggccatgtctgggcatctccatctgcatctccatccccccgctctctg gcggcaggccaaaggccaggagagatgtcggaactgtctggTGAACACCTCTCAGATCTCTCTACA
HTR1Du 7	HTR1D	C	G	S	S	cds	GE1162	TTGAAGGAAGG AGCCAAATG	TGTAGGAGATC TGAGAGGTGTT CA	TTGAAGGAAGGAGCCAAATGTgtggagggtctgtggaagagagagccacccatgcatgtccccact gaaccagtcagcagagaggccctcccccaggagccctccacacagatccctgaatgccacagaaacct cagaggcttgggataccaggaacctccaggcgctcaagatctcccttgcgtgggtcccttccgtc atcacactggccacagtcctctccaatgcttctactcaccacacatcttactcaccaggaagct ccacacccctgccaaactaactgattgtctggctccctggccacccaccccttggtttccatcttgg taatggccatcagcatcgctataccatcaccacacacccctggaaactttggccaaatcttgggtgac atctggctgtc(c/g)ctgacatcacgtgctgacagccctccatctctgtgtcatctg ctctggacaggtactgggcaatcacagatgcccctggaatacagtaaacgcagagcggtggccac gcggccacacatgctggccatgtctgggcatctccatctgcatctccatccccccgctctctg gcggcaggccaaaggccaggagagatgtcggaactgtctggTGAACACCTCTCAGATCTCTCTACA
HTR1Du 8	HTR1D	A	C	A	A	cds	GE1162	TTGAAGGAAGG AGCCAAATG	TGTAGGAGATC TGAGAGGTGTT CA	TTGAAGGAAGGAGCCAAATGTgtggagggtctgtggaagagagagccacccatgcatgtccccact gaaccagtcagcagagaggccctcccccaggagccctccacacagatccctgaatgccacagaaacct cagaggcttgggataccaggaacctccaggcgctcaagatctcccttgcgtgggtcccttccgtc atcacactggccacagtcctctccaatgcttctactcaccacacatcttactcaccaggaagct ccacacccctgccaaactaactgattgtctggctccctggccacccaccccttggtttccatcttgg taatggccatcagcatcgctataccatcaccacacacccctggaaactttggccaaatcttgggtgac atctggctgtctctgacatcacgtgctgacagccctccatctctgtgtcatctg ggacaggtactgggc(a/c)atcacagatgcccctggaatacagtaaacgcagagcggtggccac gcggccacacatgctggccatgtctgggcatctccatctgcatctccatccccccgctctctg gcggcaggccaaaggccaggagagatgtcggaactgtctggTGAACACCTCTCAGATCTCTCTACA

FIG. 5UUUU

[illegible]

FIG. 5VVV

[illegible]

FIG. 5W.WWW

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
HTRIE1	HTRIE1	G	T	E	D	cds	GE1156	GCACGACCATTGTTATCTACACC	CCCATCCACTC ATGAGGCT	GCACGACCATTGTTATCTACACCattactccagctgggtgctgtttatatcccccttgactttgactgattctctattaccggtattaccacggcgcccaagagcctttaccagaaaggggatacaagtcggaacttaagcaacagacacagatagccagaattctttgcaagttgtaaaacttacacagac ttctgtgtgtgtgacttctccactcagacccctaccacagagtttgaaaagttccatgctccca tcaggatcccccttcgacaatgatcaccacggagaaagctcagcagatctctagcacc aggaaacggagggcagcagcagctctggggctgattctgggtgcatcttattcctggctgccc attttcatcaaga(g/t)ttgattgtgggtctgagcattacacccgtgctctgggaagtggcc gacttctgagctggctgtggaattctctgataccctctgctctatacagagtttttaa tgaagactttaagctggcttttaaaagctcattagatgcgagagatactttagactgtaaaaa gctaaaaggcagcagcttttccagAGCCTCATGATGGATGGG
HTRIE3	HTRIE3	C	A	L	I	cds	GE1156	GCACGACCATTGTTATCTACACC	CCCATCCACTC ATGAGGCT	GCACGACCATTGTTATCTACACCattactccagctgggtgctgtttatatcccccttgactttgactgattctctattaccggtattaccacggcgcccaagagcctttaccagaaaggggatacaagtcggaacttaagcaacagacacagatagccagaattctttgcaagttgtaaa(c/a)ttcacac agacttctgtgtgacttctccactcagacccctaccacagagtttgaaaagttccatgccc tccatcaggatcccccttcgacaatgatcagatcacccacggagaaagctcagcagatctctag cacagggaacggagggcagcagcagctctggggctgattctgggtgcatcttattcctgggc tgccatttttcatcaagagttgattgtgggtctgagcattacacccgtgctctgggaagtggcc gacttctgagctggctgtggaattctctgataccctctgctctatacagagtttttaa tgaagactttaagctggcttttaaaagctcattagatgcgagagatactttagactgtaaaaa gctaaaaggcagcagcttttccagAGCCTCATGATGGATGGG
HTRIE4	HTRIE4	C	T	H	H	cds	GE1157	CAGCCAAAGGA AAATAACCAA	GCACCCAGCGT GGAGTAAT	CAGCCAAAGGAATAACCAacagcttctccacagctgtagactgaaacaagggaacatgaacat cacaactgtaccacagagggccagatggctataagacccaagaccatcactgagaagatgctcca tttgcatgactctgggtgctatcacacccctcacacgcttgctgaaactggctggatcatggct attggcacccaacgaagctccacacgctggccaactcaattgtctctggccgtgacgga cctcctgggtgagctgctgtcatgccccctggagcatcattacattgcatggatgctgggaagc ttgggtacttctctgtgaggtgtgggtgagtgaggacacccgtgacacccgtccatcctc cacctctgtgcatgtccctggacaggtactggggccatcaccaatggtattgaaacggcaggaa gaggacggcgaagggcggcgtgattgactctacccgtgagccatctccatttccatctcca tgccccctctgttcttggaagaccccgccgctaaagccctcccccttagctcagtgccaccatccag ca(c/t)gacatgttatctacacaaTTTACTCCACGGTGGGTGC
HTRIE5	HTRIE5	C	T	S	F	cds	GE1156	GCACGACCATTGTTATCTACACC	CCCATCCACTC ATGAGGCT	GCACGACCATTGTTATCTACACCattactccagctgggtgctgtttatatcccccttgactttgactgattctctattaccggtattaccacggcgcccaagagcctttaccagaaaggggatacaagtcggaacttaagcaacagacacagatagccagaattctttgcaagttgtaaaacttacacagac ttctgtgtgtgtgacttctccactcagacccctaccacagagtttgaaaagttccatgctctc/ t)catcaggatcccccttcgacaatgatctagatcacccagggagaaagctcagcagatctctag cacagggaacggagggcagcagcagctctggggctgattctgggtgcatcttattcctgggc tgccatttttcatcaaaagattgattgtgggtctgagcattacacccgtgctctgggaagtggcc gacttctgagctggctgtggaattctctgataccctctgctctatacagagtttttaa tgaagactttaagctggcttttaaaagctcattagatgcgagagatactttagactgtaaaaa gctaaaaggcagcagcttttccagAGCCTCATGATGGATGGG

FIG. 5XXXX

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
HTR2Ad 14	HTR2A	C	T	-	-	noncoding	GE1167	TGGTACTGCGA AACCA	CGCAGCTGCTAG GATCCTGTT	TGGTACTGCGAAGCAACCACTTATCTCTACCATGTGGGTTTGAATAATATCTGGTGGCATA TTCTGCTGAAGAAATAAGCCAGTCTCAATGTGTATCTATTAATAAATAAGTGTCTAATAGTT TATCAGAGTTATCACACAGACTGCTAGCCACCTGAGCCTATGTGGCCAAATGTCAATAATCC ACTGTGACACAAACACTGTGGCTTGGATGGAGTGGCAGACACTCACAGCACTC/T GAG GACATACTGTTTCTAGCCTTATTTATGTTCTCTCTCAACCTCAAGCCTAAAAATTACCACAG TGCCACTTACCTACCTTAATGGAAATCTGCACAAAGGCCATTTCTCCAGTTCTCTCAAAAGCA AAGGAAACCTTCCCAATATTAATGTAGCAAAAGGGGAGAGAAAGCCTGTCTGGTCCGC CCTCCTGGCTGTGCTACCTTACCTTATGACATACATAGAGGGGCTGTGATGAATGAAC GAGACAGTCAGAGAGTACTCCATCCCTGGAAACCAAGGAGTCCCTGTGGCAGACAGCTCTTC CTACTTCCCATGCACTCTTTTGTGGACTTGGGGGCTGTGAATGATTTCTAAATGTGTGC CTGCTGAGGCGAGCCGACAGAGGGAGGAGAACCCAGCCGAGCCGTGCGAGAGGAAGCCACAGG ATCCTAGCAGTGCG
HTR2Ad 15	HTR2A	G	A	-	-	noncoding	GE1169	CCGTGCCAGAG GAAGCC	CTCACCAACCC GAGGACANA	CCGTGCCAGAGAGCAACAGGATCCTAGCAGTGGGACGTGGCTCAGCTCTTGCATGCAGTTT TTGAAGTCAGCAAAACAGAAACCAATTAATCTATCTATCTGCTGGTGGAGATCAAGAGAGGG GACTCTACACCACTTAAATTAATCTGTAGAGATGCAGGAGTCAAGAAATACAAATGTATCTCAT GTGTGAACCTGAAGCAAAATGTAGTCTCATCGCTATATTTATGTGTGTAAATTTCTT TCCGGTTTGAATCATGCTTGGCCAACTGAATCAATTTCAATGAGAAATCCAGGGGAGAAAGT TGCTGCTAATCTTACTTAAGACTTTTGTCTTCTTATAGCTAAGCAACATATAGGAG CTGAAATCTCTGACAGCAGCTGTGGCAATTCAGCCCTAAGAAATGGCTGAGAACTGTAAACCAAGA TACATCCAATTAATGATTAACACTGATGATTTTAACTGACTTCTTAATGTAGAAATGT GTACATCCCCACTGTTCTGTGATGCTATTTTAAATAATACTGTGTCTAACTAGTACCATC/ G/A GCATAACCAAAATGAGATATGTTAAACAGAGTCCAGTAGTTATAAACTTTCTT CTTGTCCAGAACTTATCTTCCCGAAGCTCAAAAAAACCCTGCAACCTCTATGTATAAAG TTCTCATCTCTGCTTTTGTGCTCTGGTTTGGTGAG
HTR2Au 1	HTR2A	G	C	-	-	noncoding	GE1167	TGGTACTGCGA AACCA	CGCAGCTGCTAG GATCCTGTT	TGGTACTGCGAAGCAACCACTTATCTCTACCATGTGGGTTTGAATAATATCTGGTGGCATA TTCTGCTGAAGAAATAAGCCAGTCTCAATGTGTATCTATTAATAAATAAGTGTCTAATAGTT TATCAGAGTTATCACACAGACTGCTAGCCACCTGAGCCTATGTGGCCAAATGTCAATAATCC TTCCACTCTGGACACAAACACTGTGGCTTGGATGGAGTGGCAGACACTCACAGCACTCCGAG GACATACTGTTTCTAGCCTTATTTATGTTCTCTCTCAACCTCAAGCCTAAAAATTACCACAG TGCCACTTACCTACCTTAATGGAAATCTGCACAAAGGCCATTTCTCCAGTTCTCTCAAAAGCA AAGGAAACCTTCCCAATATTAATGTAGCAAAAGGGGAGAGAAAGCCTGTCTGGTCCGC CCTCCTGGCTGTGCTACCTTACCTTATGACATACATAGAGGGGCTGTGATGAATGAAC GAGACAGTCAGAGAGTACTCCATCCCTGGAAACCAAGGAGTCCCTGTGGCAGACAGCTCTTC CTACTTCCCATGCACTCTTTTGTGGACTTGGGGGCTGTGAATGATTTCTAAATGTGTGC CTGCTGAGGCGAGCCGACAGAGGGAGGAGAACCCAGCCGAGCCGTGCGAGAGGAAGCCACAGG ATCCTAGCAGTGCG

FIG. 5ZZZZ

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
HTR2Au10	HTR2A	C	T	A	V	cds	GE1176	TGTTTCATTTT CTGTTCAACTC C	TCACACACAGC TCACCTT	TGTTTCATTTTCTGTTCAACTCCaggtatataatgccaatcaccagctcttttgggtctacaggacga ttcgagggtctttaaggaggaggagttgcttactcgcgcgatgataaacttgttctgatcggtctctt ttgttcattcttaaccttaacctatcatgtgatcactacttcttaactaactaatcaagtccactc cagaagaagctactttgtgttaagtgtatcttggcacagggtccaaattagcttcttccagctt cctccctcagagttcttcttcagaaaagctctccagggtgcgacataggaggcagggt cctacacaggcaggaggactatgcagtcctcagcaatgagcaaaaaggcatgcaagggtgctgggc atcgctcttctctgttgggtgagtggtgctcttctcatcagaacatcatcgccgtcat ctgcaaaaggctcgcaatgaggatgtcattggggccctctcctaatgtgttggatcggtt atctctctcagcagtcacccactagctctacacactgttcaacaagacttataggtcagccctt tcaagggtatattcagtgccagtcacaggaacacaaaacacattgcagttcaatttcagtgaaacac aataccggctttggcctacaagtcagccaaactcaaatgggacaaaaggaaatccaaagcaag atgccaaagacacagataatgactgctcaatgggttg[c/t]ctaggaaagcagcatctctgaaga ggcttctaaagacaatagcgacggagtgaaatgaaggaaggtgagctggtggtg	832
HTR2Au11	HTR2A	G	T	G	*	cds	GE1172	TGCAACTCTA TGCTAAAG	TGCTCTTTATT ACCACTGCG	TGCAACTCTATGCTAAAGTtctattctgcttttttggctctcggtttgggtgagaaaaataataaa accaaacagtggaactctctctaaaatttgttaagaaagaaacttacagccacacagttcagttct ttaatcatcattgtaataatggaagcaaaaataccagccccgggagacacagcatgtacacacggcc tcagtgttacagagtggtgggtacatacagggtgaatagtgagcagaacctataacctgttagtctt tctacacctcatctgctacaagttctggcttagacatggatatcttcttggaaagaaaatactctt ttgagctcaactacgaactccctaatgcaattcaaatgatgacacacaggtctacagtaaatgactt taactcc[g/t]gagaagctaacactcttgatgcatatttaactggacagtcgacttgaaaaatcga accaaccttctctgtgaagggtgctctcaccgtctgtgtctctctactctactctccaggaaaa aaactgggtctgttactgacagccgttagtgatattcttaactatgtctggaaacatactctca tcattggcagtgctccctagagaaaagctgcagaatgccacaactatctcctgatgtcacttgcc atagctgatatgctgtgggtttctcttgctcagtcggctgtccatgtttaaccatctctgtatgtga gtggcattagtttcccagctatatctccacttggtaataataagca	750
HTR2Au12	HTR2A	C	T	-	-	noncoding	GE1169	CCGTGCACAG GAAGCC	CTCACCAAACC GAGGACAAA	CCGTGCCAGAGAGCCacaggatcctagcagtgcggaagtggtcagctcttgcacgagttt ttgaagtcagcaaaacaaacaaacttactatcatatctgtctgggagatcaagaagagg gactctacacacaggtttaaattactgtgagagatgcagcgatcacagaataacaaatgtatctcat gtggaacccctgaagacaatgtaagttctcatgccgtcatattttgtgtgtaatttctt tccggtttgaaatcatgcttggccaacatgtaacttcaatgagaatttccaggaggaaagt tgctgttaactcttacttaagactttttgttttctcttattagctcaagcaactatataagg ctgaaattcttcgacagcagctgtggcaattcagc[c/t]caagaatgcttgagaactgttaacc aagatcacatccaattactatggataaacactggatgtatttttaattgacttcttaattgaga atggtgacatccccactgtttctgattgcatgctatttttaataataactgttgcataactagacc atcggcataacaaacaaatgagatagtttaacaagaggtcccgagtagttataaaactttctt cttggctccagaaacttactctcccgaacgctcaaaaaaacccctgcacacctctatgctataaag cttgcattctgcttttttggctctgggttgggtgag	749

FIG. 5A AAAA

FIG. 5BBBBB

[illegible]

FIG. 5DDDDD

[illegible]

FIG. 5EEEEE

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
HTR5au	HTR5A	C	T	I	I	cds	GE1319	TCTGCAAGTAC CCCAGG	GGATTGCTGAG ATACCCA	TTCTGCAAGTACCCAGGCGGgtctctctgaccagagatggatttaccagtgaaactaaccctctt1800 tctcttccacccccctctcttggagacaaacacagcctcgggaaagacgacctgccccca gctgccccctgctctgggtctctggagtgctattcttacccttggggcttcttggggcg acgttcgcttggaacctgctgggtgctgggacacacctctcgtgtacgacaccttccacccgctgccc ccaaacctgggtggcatccatggcctctcggtatgtctctgggtggcgctgctgcatgccccga gctgggtgcatgagctgctggggcgccgctggcagctaggctggagagctgtgcaagcttggatc gggtgagagctgttgggtgacggccagcatctggaaagctgacggccatagccctggaccgcta ctggctccatccagcgccacatggaatacacgctccgaccccaagtgcgcttccaaactcatga tcgggctcacctgggacctctccgtgtcat[c/t]tcttggcccgctgcttcttgggtggg agagagctactctgagggcagcgagagtgccaggttaagcggagagcttctacgcccgtgtct ccacgtgagcgcttctacctggctgtgtgtgtgtgtctctggtactggaagatctacaag gctgccaagctccgggtggggtccagggaagacaaatagtggtctcaccatataccgaagctgtgga ggTGGGTATCTCAGCAATCC
HTR6d5	HTR6	G	A	P	P	cds	GE1148	CCAGCCGCTGT CGGACT	AGGGTCTGGGT TCTGCTCA	CCAGCCGCTGTGCGACTgcacatccccaggcctcttcgatgtcctcacatggctggggttactgta511 acagcaccaagaacccccatcatctaccacctcttcagtcgggagcttcaagcggcgctgggcagg tctctggcatgtccacgtgtccccgggagcgccaggccctggctctgcacatcactgctgacac ctctcagcgcccgcccgccgtcttagctacagcaggtgtctgcgctgccccctgccccg actcagattcgggactcagaacgaggttcaggcgctctctgggctcgggtcagcgcccgctg ctgcttctggcgaggccacccaggaaccccgctgcccacagggcgctgcccgcgtcaattt cttcaacatcgacccccggagcccgagctcgggcc[g/a]catcacttggctcccccaagac tgacccgggctggggtggccaatggggagctggatTCAGCAGAACCCAGACCCCT
HTR6u1	HTR6	C	A	Q	K	cds	GE1148	CCAGCCGCTGT CGGACT	AGGGTCTGGGT TCTGCTCA	CCAGCCGCTGTGCGACTgcacatccccaggcctcttcgatgtcctcacatggctggggttactgta511 acagcaccaagaacccccatcatctaccacctcttcagtcgggagcttcaagcggcgctgggcagg tctctggcatgtccacgtgtccccgggagcgccaggccctggctctgcacatcactgctgacac ctctcagcgcccgcccgccgtcttagctacagcaggtgtctgcgctgccccctgccccg cggactcagattcgggactcagaacgaggtcaggcgctctctgggctcgggtcagcgccca gctgtgcttctggagggccacccaggaaccccccgctggccacagggcgctgcccgcgtca attcttcaacatcgacccccggagcccgagctcgcgccgcatcacttggcatcccccaagac tgacccgggctggggtggccaatggggagctggatTCAGCAGAACCCAGACCCCT
HTR6u2	HTR6	G	T	A	A	cds	GE1316	GTCTCAGGA CGGTCCCC	CTCCGAGCGCT GACTGG	GTCTCAGGACGGTCCCGctccagcctgagcttcgcccggggccctcatctgcttccccgccact1807 ctatcactccttgcgctccacacctcgttctcatgttcccagagcgggcccaaccgccaatag caccggcctggggggcaggggcgccgctggcccccggggggcagggtgggtggcgccgccc tggtgctggtcacatcgcgctgacggggc[g/t]gccaaactcgtgtgatcgcgctcatctgac tcagccccgctgcgcaacacgtccaaacttctctgggtgtcgtctcactgtcagacctgatgg tggggctgggtgatggcgccggccatgctgaacggctgtacgggcgctgggtgctggcgcc ggcctctgctcttggaacccctcgacgtgatgtgtgacgacctccactccactcctg cctcatcagcctggaacgctacactgctcatctctgcgctgctgctacaagctgagcatgagc ccctgctgcccctggccctagtctggggcctggagcctgcccgtctcgtctctctgccc ctgctgtgggtggcagagctggggccacacgacacggccacccgctcctggccagtgccgctgct ggcagacctgcttctcttggcgctgggctcagctcacttcttctggcctgggtggcctat gctcactcactgcaggatcctgtgagctgccccgaagagggcgctgcagggtggcctcccccaac acccgcatgcCAGTCCGGCTCGGAG

FIG. 5FFFF

[illegible]

FIG. 5GGGG

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
IGF1d3 6	IGF1	C	T	C	C	cds	GE591	GAAAGCAGATT GCACCTAAC	CCACCCAGGTG GGCTTA	GAAAGCAGATTGACACCTTAACATgagggccactctgtttgatttggcagacacacccacagggta tggctccagcagtcggggggcgcctcagacagggcatcggtgatgagtc/c/ttgcctccggagc tggatctaaaggagctggagatgattgcgacccctcagcctcgaagctcgaagtcgctctgt ccgtgccagggccacccgacaagcccaagacccagaggTAAGCCACCTGGGTGG	253
IGF1d3 7	IGF1	C	T	-	-	noncoding	GE678	AGGGCCCTTGA GTTGCT	GCTGGATAATT CATTTGTTCTAA T	AGGGCGCTTGAGTTGCTGagatgcaggaattctataaataaccctcatagctagtaga ttggatgaattgaatgctcctgacatctcagttcttgcagtgagctatccaaataaactggccaa ctggttggtaaaagctcagctcaatctcttaaaacactttcaaaatattgtgggaagcatttg atttccaatttgattttgaaattctgcatttggttttatatacaaaagataagtgaaaagagaga aaggaataaagaaaggaataaagagattctaccagtgaaaggggaaatttaattactcttt gttag(c/t)actcactgactctctatgcagttactacataatctagtaaaaccttgtttaatac tataataatattctattctttgaaacacacatgattctcttcttctaggaataataagga aagtgcacccaaatttgaataatttaaaataataatctaaataaagtcacaaagtattcttctta acaaactttactcttacttgcctgtatatacatatttttaaaaaagtttggtaaaatagcttg actgaggttcaagttgaagggcaaaacttccatcacaaagaaatttcccatgcctgtcaga aggttagccctagctctctgtgaatgtgttttccatcactgaataatttggtatcaagaag tccactgggttagtgcactgctccatcatagcctagaaaatgatccctatctgcagatcaagattt tctcattTAGAACAAATGAATTATCCAGC	807
IGF1u1	IGF1	C	A	A	D	cds	GE638	GGAACCACTTG TTCTCAATGC	TTTCGGTTTC TCCATGTTTC	GGAACCACTTGTTCTCAATGCAattatttttggatgtttacagttacagcccccatctaccaac aagacacagagctctcagagaaggaagggttggccaaagacacatccagggagggaacagagga gggacagagaagcagctcgcagatcagaggaaggaagagcagagggaggagatttggagga gaatg(c/a)tgaaatgcagaggccaaaaggaataatgaaggacagaggatttaaacagacagag gcaaggatgatgagagggagcagacagcaaatgaagagcagaaaatacaaatagagggaatga agaaagtaggcctgctggagctagatgatgtgatggaaatagaaagtaaccccttttagagaat ctcgttaagAAACATGGAGAAACCGGAAA	419
IGF1u1 0	IGF1	A	G	-	-	noncoding	GE688	TCATAGCCTAG AAAATGATCCC TAT	AGGGTTTGAT CAATTTGTT	TCATAGCCTAGAAAATGATCCCTATctgcagatcaagatttctcattagaacaatgaattatcc agcattcagatcttctcagctcacccttagaacttttggtaaaagtcacccaggttgattatttc atgcaaatctctatatatttctacattctctggaagcttatatgaataaacaataaataatctcagtt ttctccactgggtcactcactcaagatcagagccaggaataaataaagactccctggatctc tgaatatatgcaaaaagagggcccaatttagtgagccagcaatccctgttcagtaacacagattt ttaactctcagtcacacattatttgaaattgagccctcagcagatgcttagcaatgttctaatcac tatggacagatgtaaaagaaactatacatcttttgcctctgcctgttttccagacacacacagg ttctgtggataagatactggactcctcttcccaagatggcactcttatttcttctgtcccca gtgtgtaccttttataattctcctctcaacaaactttataggcagctctctcagacttaac ta/g/tgttttctgtcagtagtgatgataattctgaagtgctcagcttatttctcctcac ttaattctatccacagtcataaaatcccccaaggaggaaagctgaaagatgcaacgtcccaatatta tctttcttaacttttccacacataatcctcctcaactgattataataaataaataaact cattataaccaattcactattttatttttttaattgaattaaactagaAAACAAATGATGCAACCC CT	847

FIG. 5HHHHH

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
IGF1u1	IGF1	T	G	-	-	noncoding	GE682	CCACACATAA TCCTCTCCAAC T	CACATTGGCAT AGCTGGC	CCACACATATCTCTCCAACTGgattataataataatgaaataaactcattattataccaattcac tattttatttttaataatgaaataaacttagaaacaaatgtagcaaaccttggaagtcagttgat [t/g]actatactacagcagaatgactcagatttcattagaaaggagcaaccaaatagtccaaa ccaaactttccaaagctttgcttcgaattgattgctttataattcttgaatgaggcaattcca agataattgtaaaagacagtaaacatttggtagaatgagcttcaactcattagcttatttcca atttaattgacatactgatacttaggtcaaatcttctctctctctcttgcacaaataataataa gtattatttgaaactttttaagatggggcagttccccctgaaaaagttaatgcagctctccatcaga atccactcttctaggatataaaaatctcttaacacccactacatacacagacacacacacac acacacacacacacacacacacacattccaccttaaggatactggaatggaatggaagaaa tcacttccctgaaaaattttattaaaaacaaacaaacaaacaaacaaacaaacaaacaaacaa tccttccctctcttggaacgtcaatgtttgttagatgaacacatctcatctgttggctccag ggttctgttactattttatgcacttgggagaaggcttagaataaaagatgttagacattttgct ttccattttattgtttggcagctatgccaattg	815
IGF1u1 2	IGF1	T	C	-	-	noncoding	GE683	TTTTATATTAC TGAGGCTAAA AGT	AGATATACCAT TTTATTATGAC ACTCT	TTTTATATTACTAGGCGCTAAAAGTaaacatttactcatttttttccccaaaaatggcactgaagt aaagtaggaaaaataaaaacagagctctaaaatcccccttcaagccacccattgacccactcac aacctatagcaaaagtcacttctgttaactcccccttaattctgattttgtttggataattattctgtac ccgctgctaaacacacactgcaggaggagactctgaacccctcaactgtctacttactcttttct gtctgtgtatcatgaaaaatgtctattccaaatatcaaaaccccttcaaatatacgcgcagcttat attcagtttcacataaaggcccccaataccactgtcagattcttttggtaaaagagttaatgaacta tgagaatttgggattacatactgtattttgectcattgtatttttatcacactttagggccaagt t/c]gataaaataaaactacagacactgaattaatttccccctgctacttgaacacagaaaaataat gactggccattcgtttacatctgtcttagttgaaaagcatttttttataaaattaatctgtatg tatttgaattattattcaattcacttattgagagggaataatcaatcctaattgactctcaaaat gtaactaaattgaatcattatcttcaatttactgttttaataagcatttttgaataatgtatggcta GAGTCTCATATAATAAATGGTATATCT	741
IGF1u1 3	IGF1	G	A	G	E	cds	GE642	AAAAATGCTTC TGTGCTCTAGT	GAATTCGCCAA TGACTTCAA	AAAATGCTTCYCTGCTTAGTTtttaaaatgcaaaaggtatgatttttttgcaccatgccccaaa aaagtccttactcaaataaactttgcagaagaggagagagagagagagagagagagagagagagag gttccctgtctacagtgctgtgtttttagataaaatgtgaggattttcttcaatcccccttctt gtttgtctaaatctcactgtcactgctaaattccagagcagantagagcctgcgcaatgggaataaag tccctcaaaattgaaaatgtgacattgtctcaacatctccatctctctcggattttctttgtcttc attattcctgtctaaccaattcatttccagactttgtacttcaagaagaatgg[g/a]aaaaatca gcagctttccaaaccccaattattttaagtgctgtctttgtgattttcttgaaggttaaatatttctac tctttgaagtcatttggggaaattc	478

FIG. 5

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
IGF1u1 7	IGF1	A	T	-	-	noncoding	GE1192	GGCTTAATAAAA ATAGCATTAGG T	GATGCCATTGC ATAAATCAGA	GGCTTAATAAAAATAGCATTAGCTCTATCTAGCCACCACCACCTTCAACTTTTATCACTCACAA gtagtactgttcaccaaattgtggaatttgggggtgagggcaggaggttggaaatttttttaaa gttagaaggctccattgttttgggtggtcctcaaaccttagcaaaatttagcaatatattatccaatc ttctgaacttgatacagagcatggagaaataaacgagggaagaaagatcttataggcaaatagaag aatttaaaagataagtaagtctcttattgatttttggcactgtctctaaaacagatatctcagc aagtggagaaataaagaaacaaagagaaataacatagatttaccctgcaaaatagcttctgccc aaantccccntggggaatccctggcaatttactggtttatagaagacattctccctcaccag acatctcaagagcagtagctctcatgaaagcaatcactgataaagaaatttgggaaattgttgaaag gtatttctcttatgagatgggggttatctactgataaagaaatttatgagaaattgttgaaag agatggctaaacaatctgtgaagatttltgtttctgtgtgtgtgtgtgtgtgtgtgtgtgtgtgt [a/t]acagctcttatgaatttcttaalgcttcaaaatgactgggtctcttcttcttcttcttctt tatcagatgaggaataaataagtttaaacccacacatagactctttaaactataggctagatagaaa tgtatgttgacttggtgaagctataatcagactattaaaatgttttggctatttttaactcttaa aagattgtgctaatatttagagcagaacctgtttgtgtctctcagagaagaagaatcttccat tcaaatcacatggcttccccaataatttcaaaagataaaTCGATTATTGCAATGGCATC	972
IGF1u1 8	IGF1	G	A	-	-	noncoding	GE683	TTTATATTAC TGAGGCTTAAA AGT	AGATATACCAT TTTATATTGAC ACTCT	TTTATATTACTAGGCTTAAAGTAAACattactcttatttggcccaaatgcactgatgt aaagtggaaataaataaaacagagctcttaaaatccctttcaagccacccattgaccccaactcac aactcatagcaaaagcactctctgttaattcccttaactctgatttgggttggatatttattctgtac ccgtgtcaaacacactgcagggaggactctgaaacctcaa [g/a]ctgtctacttactactctttt atctgtgtctgtgtatcatgaaatgtctatttcaaaatatacaaaccttttcaaatatcacgcagc ttatatctagtttacaataaaggcccccaataaccatgcagatcttttttggaaaagagtttaata actatggaattgggattacatcatgtatttggctctcatgtatttttatcacacttataggccaa gtgtgataaataaacttacagacactgaatttaattccctctgactcttgaaaccagaaataaat gactggccattctgttacctctgtcttagtgaaagacataatttttattaaatttaattctgtattg tatttgaaattatttcaattcacttaaggcagagaataatcctaatgactcttcaaaat gtaactaatgaaatcattatcttactgtttactgttttaataagcataatttggaaaatgtatggctA GAGTGTCTATAATAAATGGTATATCT	741
IGF1u1 9	IGF1	C	G	-	-	noncoding	GE1191	TTTATAGGAG TACATTGGAAG AAC	ACAACTACAA AATAGCACCAT	TTTATAGGAAGTACATTGGAAGACGcaagtagaggagtagaggagtagaggagtagaggagtag aggaaagccctcctcctgaggagtagaggagtagaggagtagaggagtagaggagtagaggagtag acctgttaaaacttgggaacacactaccacaaataaagtgtgataacattttaaagatgggggtttc ccccaatgaaataacacaaagtaaacattcccaacattgtctttaggtgatttgcaccttgcaaaa atggtcctggagttggttagattgt aaatatatatatatatatattttagtccctgcctctcaagagccacaaatgcatgggtgtgtgt atagatccagttgcaactaaatt [c/g] ctctctgaaatttggctgtggagccattctatcagca acctgtctcaagttgttatgaattgttcttatttggcacttcttctacacaaactcgggctgt ttgttttacagtgtctgataatcttgttagctataaccacacaccccttcaaacctttatat ttggcgaatttggccctcctcaagcagcagcaagtcgtcaagagcacacccaatttcaaccac aagattccatctgtgtgcatcttgaccacaaataaagtggatgcattttatattagacacaaagct ttatttttccacatcatgtcttacaataaagaaataatgcaaatagttgcaactttgaggccaatca tttttaggcataatgttttaacatagaagtttcttcaactcaaaagagttcttcaaatgataga gttaagtgcacacctaattagtaacttctcttcttcttcttcttcttcttcttcttcttcttctt tttagcatatcaattatcacaggatataatcaacagtagtgaacctctgtttctttagtataATGG TGCTATTTTGTAGTTTGT	993

FIG. 5KKKKK

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
IGF1u2	IGF1	T	A	-	-	noncoding	GE1191	TTTATAGGAAG TACATTGGAAG AAC	ACAAACTACAA AATAGCACCAT	TTTATAGGAAGTACATTGGAAGAACGcaagtagaggagggagtcgagggaacacgaagaaactacaggatgt aggaaagacccctcctgaggagtgagagagtgacatgccacgcaggatccttctgctgcacagagtt accgtttaaactttggaacacccacaaaataaagtttgataaacattttaaagatgggcgtttc ccccaatgaaatacacagaataaacattccaaacttgcttaggagtgattgcaaccttgcaaaa atggctcctggagttggtagattgctgttgaatttatacaataatgcttatagaaagaaaaa aaataataataataataataatacttagtccctgcctcctcaagagcccaaaaatgcatgggtgtgt atagatccagttgcataaattccctcctgaatttggctgctggagcattcattcagcaacct tgttaagtggtttatagaaattgtttccttatatgccaccttcttccacaaactggggtgttgt tttacagtgctcgataaattgtgtgtgtctatacccaacctcctccataaccttatattgc cgaatttggcctcctcaagagcagcagtcgcaagagcagcccaaatcccaacacaga ttccatctgtggcatttgcacaaa[t/a]ataagttggatgcatatttatattagacacaaaagct ttattttccacatcatgcttacaaaaaagaataatgcaaatagttgcaacttgaggccaatca tttttaggcataatgttttaaacatagaaaagtttcttcaactcaaaaagagttccttcaaatgatga gttaatgtgcaacctaaatagtaacttctcttttatttttccatatagagcactatgtaaa tttagcatacaattatcacaggatataatcaaacagtagtaaaaactctgttttttagtatataATGG TGCTATTTTGTAGTTGT
IGF1u2	IGF1	T	C	-	-	noncoding	GE1191	TTTATAGGAAG TACATTGGAAG AAC	ACAAACTACAA AATAGCACCAT	TTTATAGGAAGTACATTGGAAGAACGcaagtagaggagggagtcgagggaacacgaagaaactacaggatgt aggaaagacccctcctgaggagtgagagagtgacatgccacgcaggatccttctgctgcacagagtt accgtttaaactttggaacacccacaaaataaagtttgataaacattttaaagatgggcgtttc ccccaatgaaatacacagaataaacattccaaacttgcttaggagtgattgcaaccttgcaaaa atggctcctggagttggtagattgctgttgaatttatacaataatgttctatagaaagaaaaa aaataataataataataatacttagtccctgcctcctcaagagcccaaaaatgcatgggtgtgt atagatccagttgcataaattccctcctgaatttggctgctggagcattcattcagcaacct tgttcaagtggtttatgaattgtttccttatattgcaacttcttccacaaactggggtgttgt ttcacagtgctcgataaattgtgtgtctatacccaacctcctcccttataaccttatattgc cgaatttggcctcctcaagagcagcagtcgcaagagcagcccaaatcccaacacaga ttccatctgtggcatttgcacaaaataaagttggatgcatatttatattagacacaaagctttat ttttccacatcatgcttacaaaaaagaataatgcaaatagttgcaacttgaggccaatcatttt taggcataatgttttaaacatagaaaagtttcttcaactcaaaagagttccttcaaatga[t/c]ga gttaatgtgcaacctaaatagtaacttctctttttattttttccatatagagcactatgtaaa tttagcatacaattatcacaggatataatcaaacagtagtaaaaactctgttttttagtatataATGG TGCTATTTTGTAGTTGT
IGF1u2	IGF1	T	G	-	-	noncoding	GE683	TTTTATATTAC TGAGGCGCTAAA AGT	AGATATACCAT TTTATTATGAC ACTCT	TTTTATATTACTGAGGCGCTAAAAGTaaacattactcattttattttgccccaaaaatggcactgatgt aaagtaggaaaaataaaaacagagctctaaaaatcccttccagcccccatttgccccactcacc aacctatagcaaaagtcacttctgttaactcccttaactgtatttggttgatatattcttctgtac ccgtctgctaaacacactgagggaggagctctgaaacctcagctgtctacttacaatttttatct gtgctgtgtatcatgaaaatgtctattcaaaatatcaaacctttcaaatatcacgcagcttat attcagtttacaataaggcccccaataaccatctcagatcttttttggtaaaagagtaaatgaacta tgaaattgggattacatcatgtatttgcctcatgtatttttatcacacttataggccaagtgt gataaataaacctacagacactgaaataatttccctcctgcttcttgaaccagaaaaataatgact ggccattcgtttcaatctgcttagttgaaaaagcataatttttatttaaatatttctgattgtatt tgaattattattcaattcacttaaggcagagggaatatcaactcaacttctcttaaaaaatgttaa ctaaattgaatcatta[t/g]cttaccatttactgttttaataagccataattttgaaaaatgtatggcta GAGTGTCAATAAATAATGGTATATCT

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
IGF1u2	IGF1	T	G	-	-	noncoding	GE1192	GGCTTAATAAA ATAGCATTAGG T	GATGCCATTGC ATAAATCAGA	GGCTTAATAAATAGCATTAGGTCATCTATCTAGCCACCACACACCTTTCAACTTTTATCAGCTCAGCTGAGTGTCTCACCAGATTTGTAATTTGGGGTGCAGGGGCGAGGAGTTGGAAATTTTCAAA GTAGAAGGCTCCATTGTTTGTGTGCTCTCAAACTAGCAAAATAGCAATATAT/t/gjatcc aatctctgaacttgatcaagagcatggagataaaccgggaaaaaagatcttataggcaata gaagaattttaaagataagtaagttctcttattgatttctgctcgtctcaaacacagataatt cagcaagtggagaaaaataagaacaaagagaaaaataacatagattttacctgcaaaaaatagcttc tgcacaaantccccntgggaattcttgcaatttactggtttatagaagacattctccctccac ccagacattctcaagagcagtagctctcatgaaaagcaactcactgactcatttgggaaaatgttg gaagatttctcttatgagatgggggttatctactgataagaagaatttatgagaaaattgttg aaagagatggctaaacaattctgtgagatctttttgtttcttggtttttgttttttttttttt ctttatacagctctttatgaattcttataatgttcaaaatgacttgggtctctctctctttttta tatcagaatgaggaataataaagttaaacccacacatagactctttaaactataggtatagataaaa tgtatgtttgtactgttgaagctataacagactattttaaattgttttcttatttttaacttaa aagattgtgctaaatttattagagcagaacctgtttggctctctcagaagaaagaaatcttcccat tcaaatcacatggctttccaccaatttttcaaaagataaaTCTGATTATTCATGGCATC	972
IGF1u2	IGF1	G	C	-	-	noncoding	GE1192	GGCTTAATAAA ATAGCATTAGG T	GATGCCATTGC ATAAATCAGA	GGCTTAATAAATAGCATTAGGTCATCTATCTAGCCACCACACCTTTCAACTTTTATCAGCTCAGCTGAGTGTCTCACCAGATTTGTAATTTGGGGTGCAGGGGCGAGGAGTTGGAAATTTTCAAA GTAGAAGGCTCCATTGTTTGTGTGCTCTCAAACTAGCAAAATAGCAATATATATCCCAATC ttctgaacttgatcaagagcatggagataaaccgggaaaaaagatcttataggcaaaatagaaag aatttaaaagataagtaagttctcttattgattttgtgcaactctgctctaaacagatatctcagc aagtggagaaaaataagaacaaagagaaaaataacatagatttacctgcaaaaaatagcttctgccc aaantccccntgggaattcttgcaatttactgggttatagaagcattctcccttccaccag acattctcaagagcagtagctctcatgaaagcaaatcacatgactctcatttgggaattgttgaaag gtattctcttatgagatgggggttatctactgataaaagaaatcttatggagaattgttgaaag agatggctcaacaattctgagatcttttctctctgtgtttgttttttttttttttttttttt atacagctcttatgaatttcttaaagttcaaatgacttgggtctttctttcttttttttttttt agaaatggaggaataaataagttaaaccacacatcactgactctcaaatcactgagctatagaat(g/ c)tatgttttggctgttgaagctataatcagactattttaaattgtttgtctatttttaatttaa aagattgtgctaaatttattagagcagaacctgtttggctctctcagaagaaagaaatcttccat tcaaatcacatggctttccaccaatttttcaaaagataaaTCTGATTATTCATGGCATC	972
IGF1u3	IGF1	T	C	-	-	noncoding	GE1326	GCTTATTATT CCACATCATGC	CCTAGAAAAGA AGGAATCAATTG	GCTTATTATTTCCACATCATGCTTCAAAAAAGATAATAGCAAAATAGTTGCAACTTTGAGGCCAA tcaatttttagctatgttttaaacatagaaagtctttctcaactcaaaagagtctcttcaaatga tgagttaatgtgcaaccttaatttcaactttcttttttttttttttttttttttttttttttttt aaatttagcatatcaattatcacagatataatacaacagtatgttaaactctgttttttagtataa tgggtctattttgtagtttttatatgaagagctctggcacaacggtaaatcgtgaaagcaca caatgggggaagcctggagcccaagatgacacaaaggggaaggggtactgaaacacacattctg ggaaagaaggcaagctccccagttatgcttccaagaggaacttcagacacacaaagtccactg atgcaaatggactggagctccagaggaagaaactgtgaaatggaaagagcagacagaggttagga tttagcagctctgtgttt acctgtgtgaccttggcagtagcttcaactctctctctgctcctgactttctctatctgcaaat gggggcaatatgtctcatctactactcaaaaggggttggttatgaaggtttaaaaagataaagattca atttttttaccctgggttgctgaaggttgcaacatcagggcgtttgagttgctgagatgcaagg aatctataaataaccatttcataagcatagtagagattgggtgaattgaatgctctctgacattc agttctgtcagtgagctatccaataaactggcacaactgtgtttaaagctaaacagctcaatc tcttaaaacaccttttcaaatatgtgggaagcaatttgaatttcaatttgatttttgaattctgcat tgcattttatcaatcaaaagttaattgaataaanaagaaagaaagaaagaaagaaagaaagaa	1196

FIG. 500000

[illegible]

126/178

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
IGF1u3 3	IGF1	T	A	-	-	noncoding	GE1326	GCTTTATTTT CCACATCATGC	CCTAGAAAGA AGGAATCATTG	GCTTTATTTTCCACATCATGCCTTACAAAAAAGATAATGCAATAGTTGCAACTTTGAGGCCAA TCAATTTTAGGCATATGTTTAAACATAGAAAGTTCTTCAACTCAAAAGAGTTCTCTCAAAATGA TGAGTTAATGTGCAACCAATTAATAGTAACCTTCTCTCTTTTATTTTCCATATAGGCACTATGT AAATTAGCATATCAATATATACAGATATATCAAAACAGTATGTAAACTCTGTTTCTTAGTATAA TGGTCTATTTTGTAGTTGTTATATGAAAGTCTGCCCCAAACGGTAATAACGGAAGCAAAA CAATAGGGGAGCCTGGAGCCAAAGATGACACAGGGGAGGTTACTGAAACACCATCCATTTG GGAAGAGGCAAGTCCCCCAGTTATGCTTCCAGAGGAACCTCAGACACAAAAGTCCACTG ATGCAAAATGGACTGGAGTCCAGAGGAGGAACCTGTGAAATGGAAAGCAGAGGCTAGGAAT TTAGCAGTCTGGTTCTTTTCTCATGGAAGAAATGAACA [t/a] CTGCCAGTGTGTCTCATGG ACTCACCACTGTGTGACCTTGGCAAGTCACTTCACTCTCTGTGCTCAGTTCTCTCATCTGCA AAATGGGGGCAATATGTCTACTACTACTCACTCAAAAGGGTGGTATATAAGGTTTAAAGATAAAGAT TCAGATTTTCTACCTGGTGTGCTGTAAGGTGCAACATCAGGGGCTTGGTTGCTGAGATGC AAGGAATCTATAAAATACCCATCTCATAGCATAGCTAGAGATGGTGAATGAAATGCTCTGACA TCTCAGTTCTGTGAGTGAAGCTATCCAAATCACTGAGCACTAGTTGTTAAAGGCTAACAGCTC AATCTCTAAACACTTTTCAAAATATGTGGGAAGCATTTGATTTTCAATTTGATTTTGAATCTC GCATTTGGTTTATGATACAAAGATAAGTGAAGAGAGGAGGAAAAA	1196
IGF1u3 4	IGF1	T	G	-	-	noncoding	GE176	AGATAAGACAG AGGCCCAGG	TGCGGCAAAAT AAAATGAGTAA TGT	AGATAAGACAGAGGCCAGGGGATTTTGAAGTGTCTTATCTGCCCCCATCCCAACCCAGCC CTTATTTTAGTATCTGCTCAGAAATTTTAAAGAGGTGACCAAGCTGAACTCTAGAATTA AAGGAACCTCACTGAAACATATA [t/g] TCCGTTGCTCCTCTCTCTTTTCTCTTTTGTGATG GGTCTCGCACTGCCCCAGGTGAGTGGATGATCTCGGTCACTGCAACCTCCACC TCTGGGTTTAAAGCATCTCTGCTCAGCTCAGCTCTGAGTGGATTAACAGCACCCACCAAC TAGCCCCGCTAAATTTTGGATTTTAAATAGAGACGGGTTTACCATGTTGGCCAGGTTGGAC TCAAACTCTGACCTTGTGATTTGCCGCTCAGCTCCAAATTTGCTGGGATTAACAGGCAAGG CCACACACCTGCTGCTGCTTCTCTTAAATGATGATTAACATGATCTTAAACATGATCTCT CTCTCTCAATCTCAACTATCTTGTGGGTC [t/g] TCAAGGGGAAAAAATCCCAAGCTTT TTTAAAGTAAAAAAGAGAGGAGGACACAAAAACCAATGTTACTGCTCACTGCAATATAGG TTAAGATGGAGACAGGTTCTCTCAATAACCGAGCTGATTAACCTTCACTTCAAAAAACATG ACCTTCCCAATCTCTAGAACTCTGCTCTTTTATATATTAGGCTCAAAAGTAAACATTACTC ATTTTATTTTCCCAA	796
IGF1u4	IGF1	T	G	-	-	noncoding	GE176	AGATAAGACAG AGGCCCAGG	TGCGGCAAAAT AAAATGAGTAA TGT	AGATAAGACAGAGGCCAGGGGATTTTGAAGTGTCTTATCTGCCCCCATCCCAACCCAGCC CTTATTTTAGTATCTGCTCAGAAATTTTAAAGAGGTGACCAAGCTGAACTCTAGAATTA AAGGAACCTCACTGAAACATATA [t/g] TCCGTTGCTCCTCTCTCTTTTCTCTTTTGTGATG GATGGGTTCTGCACTGCCCCAGGTGAGTGGATGATCTCGGTCACTGCAACCTC CACTCTGGGTTTAAAGCATCTCTGCTCAGCTCAGCTCTGAGTGGATTAACAGCACCCCA CCATATGCCCCGCTAAATTTTGGATTTTAAATAGAGAGGGGTTTACCATGTTGGCCAGGTT GGACTCAACTCTGACCTTGTGATTTGCCGCTCAGCTCCCAAAATGCTGGGATTACAGGCA TGAGCCACCAACCTGCTGCTTCTCTTAAATGATGATTAACATGATCTTAAACATGAT CTTCTCTCTCAATCTCTCAACTATCTTGTGGGTTCTTCAAGGGGAAAAAATCCCAAGCTTT TTTAAAGTAAAAAAGAGAGGAGGACACAAAAACCAATGTTACTGCTCACTGCAATATAGG TTAAGATGGAGACAGGTTCTCTCAATAACCGAGCTGATTAACCTTCACTTCAAAAAACATG ACCTTCCCAATCTCTAGAACTCTGCTCTTTTATATATTAGGCTCAAAAGTAAACATTACTC ATTTTATTTTCCCAA	796

FIG. 5QQQQQ

SUBSTITUTE SHEET (RULE 26)

SUBSTITUTE SHEET (RULE 26)

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
LIPC ₁ 5	LIPC	G	C	G	A	cds	GE323	CTCCCGCGTAA CCCTTACC	CGGCCCATGAC TTCAATTCTC	CTCCCGCGTAAACCTTACCCctgcttcccatattagggctggatgcccgggacacctttgtttgagg gaagtgcctccacgaatcgcttctccagatgatcccaatttctggatggccatttacccttt accgggagacacatgggctgagcgtggcgcacacacagcccataggaactatgactctatcc caacggggctctctccagctg(g/c)ctgccacttcttagagctctacagacatatggccag cacggcttcaatggcagaaatgagatgagggccg
LIPC ₁ 6	LIPC	A	T	M	L	cds	GE374	TGAGAACCAAG TGATCCTCTGA	ATCTGCTATCC TGCCCTTC	TGAGAACCAAGTATCTCTGAGttgagctgcttggcgttaaggggtgataacgctcttcttgc cctgtgttccagccatcaccagaccataaaatgttccacgagcgtgctggtgacaccttttcatc gactccttctgctgacccgacgagcag(a/t)tgccctacccgtgctggtgacatgaacagct tcagccagggccttgcctgagctgcaagagggcctgcaacacgctgggtaccacgtccgc cagggagcgggagcaagagagagggctcttctctgtaacggagcccgcccccttcaaaagg tgagtgtggagctgggagccttcaGAGGCGCAGGATGTCAGGAT
LIPC ₁ 7	LIPC	G	A	E	K	cds	GE475	AACGATTGTG TCTGATTTTCT	TGGCACAAGTG GGTGCTTA	AACTGATTGTGCTGATTTCTTtgttattcaaggggcaaaagaaattgctagtaataaaacgta ttccttttctacagctggatgtggatattcggcagctgactgactgactcaagttcaagtg(g/a)aaacagtgctggtggccaatgtctggacacaggtccagacatcatccatggagcacagg ggcgccactcagggctcttctgaagacgactcagagtcacaaagcaggagaaacccagcaagggt gactgctgattcaatctcttattacgtccatTAAGCACCACCTTGTGCCA
LIPC ₁ 8	LIPC	G	T	A	S	cds	GE346	GCTGGAGAAGG AAGAAGGTA	TCACCTCTCAGA GGAAGGAAA	GCTGGAGAAGGAAGGGTAGggggagaaagggaaactaaggcgacccctctctgccccctc ctcaggtgacggctgctgtagaaactgctgagctgagctgagcgtgagctgagctgagcgtg gcccagccagtggaagctgggtggatggatgacacccctggccacgacacacacacacacg cgtccgcaacaccccgcttctgggcaagaggtc(g/t)cggtctctctcgggtggctgaggtta ccgacctggcccgagctctctctcactcccttccctcttCCCTTCTCTGAGAGTGA
LIPC ₁ 9	LIPC	C	A	-	-	noncoding	GE323	CTCCCGCGTAA CCCTTACC	CGGCCCATGAC TTCAATTCTC	CTCCCGCGTAAACCTTACCCctgctt(c/a)ccattagggctggatgcccgggacaccttgttt gagggagtgcccccgacgaatctcttctccagatgagtcgaatttttggatggcattcatatc ctttaccgggagacatggcctgagctgggacatacaacagcccataggaactatgacttct atcccaacgggggctctctccagcctggctgacacttccctagagctctacagacatatggccag cacggcttcaatggcagaaatgagatgagggccg
LIPC ₂	LIPC	T	A	V	E	cds	GE346	GCTGGAGAAGG AAGAAGGTA	TCACCTCTCAGA GGAAGGAAA	GCTGGAGAAGGAAGGGTAGggggagaaagggaaactaaggcgacccctctctgccccctc ctcaggtgagcggctgctgtagaaactgctgagctgagctgagcgtgagctgagctgagcgtg gcccagccag(t/a)gaaagctgggtggatgagctgagctgagctgagctgagctgagctgagc tgccgtccgcaacaccccgcttctgggcaagaggtcggcgtcttctcgggtggctggaggtta ccgacctggcccgagctctctcactcccttcccttcccttCCCTTCTCTGAGAGTGA
LIPC ₂ 0	LIPC	A	T	I	F	cds	GE300	TGGGCAAGTCTT CCCTAACAA	ACCCCTGGATT CTTTGTGAC	TGGGCAAGTCTTCCCTAACAAagtatctaataggcattgtggtctcttgggttcagaaattacca agaaagcctggaccgggtgaaacgggaaatgggacaaagtccttctgttcttccattctgt tggttttatgcatctt(a/t)cccaataagtgcccttggaacaaagcctgaaacccaggttaagag cctgactttctccagagatgggcatgactttctttttaaaccgtgtGTCAAAAGAAATCCAG GGT
LIPC ₂ 1	LIPC	A	C	H	P	cds	GE253	CACACTGGACC GCAAAAGG	AGTGTGTGAGT TATTAGGCATG G	CACACTGGACCGAAAGGcttctcatccaggcagctcttctctgcccccatcccgctgctgc ttccagggaatctgttcaactctctcgaagcagctgttccactaatgggtacagcctgggtgcac a/c)cggtcaggatttgcggcagttccatcggtggaacgcaagattgggagaaatcacaggt aaCCATGCTAATAACTCACACT

FIG. 5VVVV

SUBSTITUTE SHEET (RULE 26)

[illegible]

FIG. 5YYYYY

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
MPLu4	MPL	C	G	L	V	cds	GE482	CTCTGGTGGCA CAATGCT	CCAGGGATCCC CTGGGTA	CTCTGGTGGCAATGCTTGTGACAGAGGACCTAAGCTGCTCCCTGCTGACATCCCTGtagt ggcctcccccaccccaacttgactggaggagatccagtgggcat[c/g]tggaattggag tggcagcaccatctgctctggcagcccaagagacctgttatcaactccgatacacaggagaagg ccatcaggactggagggtatggtaagcaacaatgccacagacctcacTACGACGGGATCCC TGG	263
MPLu5	MPL	G	A	E	K	cds	GE491	AGAGGCTGAGC CATAGACTGT	TGGGGCAAGAT TGAAGGTAG	AGAGGCTGAGCCTAGACTGTGtactcagagtlctgatgtgcccgtctctggccctcaggcctgc cggctccccccagtatcatcaaggccatgggtgggagcagccaggggaaacttcagatcagctgg gaggagccagctccagaaatcagtgatttctctgaggtac[g/a]aactccgctatggccacagag atcccaagaactccactggctccacgggtatcacagctgattggccacagaaacctgctgcccctgct ctgcagaggcctcactcagcctctgctctgacacagtgctccatgtgctcagcccacaatgccctg gcaagatggaccacaagcagacctcccccaagtagagagatgctgacctctctgtgccccacctc ttatctctTACCTTCAATCTTGGCCCA	417
MPLu6	MPL	C	A	R	R	cds	GE490	GGGTGGAGGC TCTCTCAG	CAGGCTTCCCT AGAGATATTCT TTTA	GGGTGGAGGCTCTCTCAGctgacagcagcagctagatltgtgaagctgggattttctccccaagg cttcagctctgacagagagggggagagctgctcctcctcagagctccagctggcaactcctac tggctgcagctgg[c/a]agcgaacctgatgggatctcctcctggctgctgggagatctctggt ccctccctgctgactggacctgctgagatgcagtgagtcacaaggaataggggagatggg gaggagatAAAGAAATATCTCTAGGGAAGCCTG	293
MPLu7	MPL	T	C	F	S	cds	GE472	ACGTGGGGCTG TATCTGACA	CAGGGCTCCCT CTTCTG	ACGTGGGGCTGTATCTGACAGgaacctgaggggctggctgggaggggattggggccagcttcc tgaaggaggatgggttaaggcagcacacagtgccgagagaagatggcctcctgggcccctct/ c]catggtaacctctcctcctcctggccctcaaaacctggcccaagtcagcagccaagggtga ggtcacacagagggtggagatcactatgcccCAGGAAGAGGGAGCCCTG	244
MPLu8	MPL	C	T	S	S	cds	GE472	ACGTGGGGCTG TATCTGACA	CAGGGCTCCCT CTTCTG	ACGTGGGGCTGTATCTGACAGgaacctgaggggctggctgggaggggattggggccagcttcc tgaaggaggatgggttaaggcagcacacagtgccgagagaagatggcctcctgggcccctctca tggtaacctc[c/t]tgccctcctggccctcaaaacctggcccaagtcagcagccaagggtga ggtcacagagggtggagatcactatgcccCAGGAAGAGGGAGCCCTG	244
MPLu9	MPL	G	A	G	G	cds	GE491	AGAGGCTGAGC CATAGACTGT	TGGGGCAAGAT TGAAGGTAG	AGAGGCTGAGCCTAGACTGTGtactcagagtlctgatgtgcccgtctctggccctcaggcctgc cggctccccccagtatcatcaaggccatgggtgg[g/a]agcagccaggggaaacttcagatcag ctggaggagccagctccagaaatcagtgatttctctgaggtacgaaactccgctatggcccccagag atcccaagaactccactggtcccacgggtcatcacagctgattggccagaaacctgctgcccctgct ctgcagaggcctcactcagcctctgctctggaccagttctccatgtgctcagcccacaatgccctg gcaagatggaccacaagcagacctcccccaagtagagagatgctgacctctctgtgccccacctc ttatctctTACCTTCAATCTTGGCCCA	417

FIG. 5ZZZZZ

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
NGFBd9	NGFB	G	A	A	A	cds	GE1185	CACATCCATAC TGCTTGAGT	AGGGCCCCAGG AGAGTG	CACATCCATACCTGCTGAGTcagcccccgggttacgctgttgcctccgggtataaccattgtagca cacccttccctctcagaagcgccttgaatgaacaccttctgtagccttgccttggagggtca actctggaggaccagaaactccttctgactgcatcttagtactccatgagtcacacctcatttc tttttcattccagggtgcatagcgttaattccatgttgccttctacactctgacacacgttttctga tcggcatcacaggcig/a gacccacactcagagagaatgtccctgcaggacacacacccccca agtccactggactaaacttcagcattcccttgacatgacctgcagagcccgagcgcctccgg cagcgggatagctgcacgcgtgggggcagaccgcgaacttactgtgaccccccaggtgttt aaaaagcggcgactccgttcccccgtgtgctgttagacacccagcctccctgtgaagctgcaga cactcagatctggacttcgaggtcggtggtgctgctcccttcaacaggacctcacaggagcaagc ggtcatcatcccatcccatcttccacagggcgaaatctcgggtgtgtagcagtgctcagctgtgg gttgggataagaccacccgcacagacatcaagggaaggaggtgatggttgggagaggtgaa cattaaacacagtgatttcaacagactcttcttggagaccagtgccgggaccccaatccccgtg acagcgggtgctggggcattgactcaaacactggaactcattgtaccacgactcacaccttt gtcaaggcgtgacccatggatggcaagcaggtgcctggcggttatccggatagatcacggcctg tctgtgtgtgctcagcaggaaggtgtgagaagagcctgacctgcgcagacagctccccctctg cccccttctcACTCTCTCTGGGCCCe
NGFBu1	NGFB	G	C	-	-	noncoding	GE1186	TTTACAGAGGA GCTGACGTTTG	GACACAGGCG TAACCG	TTTACAGAGGAGCTGACGTTTgctacacatctacagtatgcagtaggagctcccgaggagccagt gagagccctccagagcagagaactaattccacaattacttgaccaagtgggggattatttgggg gig/c taactgcagtgagtgagtgctctcttgggacagtttagagccataccatttgatcta tagtcacataagaacaaacataaaaaaagacatgcttagagtgaaagagaaagggaggga gaaaagaggaaagggtggatggaaggacactagcttagtaagggtcaacttggattctatttc tggttcagtttctcatttctgacttcagtgctttagtgtagtctcatttcttgatcagtttcc ttatatgtgaaataaataatgataaatcccaattgaactcacagactcatgagaagatagaagtga acacattttaaataacatcacaaagaggaactattatgtggtccacatttatatatgtgggga gcgtctgaagaggcctggactaagaagggtccagagccacaggtttttggccaaacatgacgc tttgatattcacaaggggtcccaagtcacagatcttagagctgacccagtgcaactgtctga aagggttaccagttctgaggttcaagacatgtccccagcagatcttccccgtgcttccaga ggattcaaaactgttggcagacggcaccatcacatcaaggacacagtgccaggaggaggtgtt aaacttcccccaaacctccctggtaacacatggacacttaccacctccctcagcgccttaa gcttcagagaaactcaaggactctgtgaagtgtgtctccagctcatatcgaaactactgggcaa atttcagggtctctgctcacttccctggagagctcgagtggtggacacacacatccatctgctg agtcagccccGGGTACGCTTGTGTC

FIG. 5A AAAAA

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
NGFBu2	NGFB	A	G	-	-	noncoding	GE1186	TTTACAGAGGCTGACGTTTG	GACAACAGGGG TAACCCG	TTTACAGAGGAGCTGACGTTTGctacacatctacaagtatgcataaggagctccgcggaggccagt gagaggccctccaggagcagaactaatccacaattacttgaccaagtgggggattatttggg ggtaactgcagtcagtagtagtgctcttggggacagcttagagccataccatttgatctatagt cacataagacaacaataaaaagaaagacatgcttaagagtgaagagaaagggaggagaaa agaaggaaagggtggatgggaagacactagcttagtaagggtcaactttggaattcttctgggt tcagtttctatttgtagcttcagtgcttttagtgtagttcatttctgtgaatcagtttcccttat atgtgaaataaataatgataaaatccaaatgaaactcacagactcatgagaagatagaagtgaacac attttaaataacatcacacaagagggaactatttatgtgtccacatttatatatgtgggttagcgt ctgaagggtgcttggaactaagatgggtccagagccacaggtttttgccaaacatgacgctttg tgaattcataaacaagggtcc(a/g) agtcaccagatcttagagctgacccagctgcactgctga aagggtgatacagttctgaggttcaagacatgtccccagcagatcttccccgtgccttcccca ggattcaaaactgttgagcaggacggcaccatcacatcaagggcacagtccctcagccgcttaa aaactctcccacacacactccctggtaacacatggacacttaccactccctcagccgcttaa gcttcagagaactcaaggactctgtaagtgtgtctccaagctcatatcgaaactactgggcaaa atttcagggtctctgacttctctggagaagctcggtgggtgaccacacacatactactgacctg agtcagcccccgggtTACGCCCTGTGTC	1002
NGFBu3	NGFB	G	T	-	-	noncoding	GE1186	TTTACAGAGGCTGACGTTTG	GACAACAGGGG TAACCCG	TTTACAGAGGAGCTGACGTTTGctacacatctacaagtatgcataaggagctccgcggaggccagt gagaggccctccaggagcagaactaatccacaattacttgaccaagtgggggattatttggg ggtaactgcagtcagtagtagtgctcttggggacagcttagagccataccatttgatctatagt cacataagacaacaataaaaagaaagacatgcttaagagtgaagagaaagggaggagaaa agaaggaaagggtggatgggaagacactagcttagtaagggtcaactttggaattctattctgggt tcagtttctatttgtagcttcagtgcttttagtgtagttcatttctgtgaatcagtttcccttat atgtgaaataaataatgataaaatccaaatgaaactcacagactcatgagaagatagaagtgaacac attttaaataacatcacacaagagggaactatttatgtgtccacatttatatatgtgggttagcgt ctgaagggtgcttggaactaagatgggtccagagccacaaaggtttttgccaacatgacgctttg tgaattcataaacaagggtccaaagtcaccagatcttagagctgacccagctgctgaaag(i g/t) ggttaccagttctgaggcttcaagacatgtcccagcagatcttccccgtgccttccccaga ggattcaaaactgttgagcaggacggcaccatcacatcaaggaagtagtgcagctgctgaaag(i aaactctcccacacacactccctggtaacacatggacacttaccactccctcagccgcttaa gcttcagagaactcaaggactctgtaagtgtgtctccaagctcatatcgaaactactgggcaaa atttcagggtctctgacttctctggagaagctcggtgggtgaccacacacatactactgacctg agtcagcccccgggtTACGCCCTGTGTC	1002

FIG. 5B BBBBBB

141/178

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
NTRK1d 10	NTRK1	C	T	-	-	noncoding	GE1290	GCTGCCCTGGG TGAACA	GGGATGTCTAT AGGGAAGGGA	GCTGCCCTGGGTAACAGcagtgaggct[c/t]ggcccccaactcagtcctgtcctccgcttc catccaggcactgaaggagcgtccgagagtgctcggcaggactccaacgtgagctgagctgc tcaccatgctgcagcaccagcacatcgcgcttcttcggcgctctgacacgaggggccgccccctg ctcatgtctcttcaggtatatacgcgacacggggacccaacgcttctcccggtaccagcacctggc ctcagcgctggccccggccccctggctctggggccccctgtTCCCTTCCCTATAGACATCCC
NTRK1d 9	NTRK1	G	C	-	-	noncoding	GE1067	GGAGGCTCTGA GAGTACAGGAG	AGTAGGAACA AAGCCAGGAG	GGAGGCTCTGAGAGTACAGGAG[g/c]agccccctggatataaactacccctgtcccccaagggtc tcgggtggctgtggggcctggccgctcttcttcctgctcttcttctacgctgctccttctgtgctcaa caaatgtggacggggaacaaagtttgggatacaacgtagtcggggctgcagagggtgtctgtct gtctgtctTCCCTGGCTTGTTCCTACT
NTRK1u 1	NTRK1	A	G	Q	Q	cds	GE1290	GCTGCCCTGGG TGAACA	GGGATGTCTAT AGGGAAGGGA	GCTGCCCTGGGTAACAGcagtgaggctggcccccaactcagtcctgtcctccgcttccttc caggcaactgaaggagcgtccgagagtgctcggcaggacttcca[a/g]cgtgagctgagctgc tcaccatgctgcagcaccagcacatcgcgcttcttcggcgctctgacacgaggggccgccccctg ctcatgtctcttcaggtatatacgcgacacggggacccaacgcttctcccggtaccagcacctggc ctcagcgctggccccggccccctggctctggggccccctgtTCCCTTCCCTATAGACATCCC
NTRK1u 2	NTRK1	C	T	F	F	cds	GE1290	GCTGCCCTGGG TGAACA	GGGATGTCTAT AGGGAAGGGA	GCTGCCCTGGGTAACAGcagtgaggctggcccccaactcagtcctgtcctccgcttccttc caggcaactgaaggagcgtccgagagtgctcggcaggacttccaacgtgagctgagctgcctcac catgctgcagcaccagcacatcgctggctctt[c/t]ggcgctgcacacgaggggccgccccctg ctcatgtctcttcaggtatatacgcgacacggggacccaacgcttctcccggtaccagcacctggc ctcagcgctggccccggccccctggctctggggccccctgtTCCCTTCCCTATAGACATCCC
NTRK1u 3	NTRK1	C	T	H	Y	cds	GE1129	CAGGCTCCTGG GAGTCTATC	CCAGGTGTCT ACAGTTGGAT	CAGGCTCCTGGGAGTCTATCctccagcctatccccctctcttcttctgttcacagatcc[c/t] latggaccagatgccaaagctgtggctgtggggagagtggtggtccagggccccctgggtctggg gcagctgtgctggcggtggctagcaggtcgctgcggggatggtgacctggcggtctgcatctttg tgacacgggacctggccacacacgactgtctagtggccaggaggtggtggtcaagattggtgat tttggcatgagcagggatatactacagcacgactattaccgtgaagggtccttgtcccccaacg ccttccccctgcATCCAAACTGTAGACACCCCTGG
NTRK1u 4	NTRK1	C	T	A	A	cds	GE1129	CAGGCTCCTGG GAGTCTATC	CCAGGTGTCT ACAGTTGGAT	CAGGCTCCTGGGAGTCTATCctccagcctatccccctctcttcttctgttcacagatcccatg gaccogaagccaaagctgtggctgtggggagagtggtggtccagggccccctgggtctggggcag ctgtggc[c/t]gtggctagccaggtcgctgcggggatggtgacctggcggtctgcatctttg tgacacgggacctggccacacacgactgtctagtggccaggaggtggtggtcaagattggtgat tttggcatgagcagggatatactacagcacgactattaccgtgaagggtccttgtcccccaacg ccttccccctgcATCCAAACTGTAGACACCCCTGG
NTRK1u 5	NTRK1	T	G	-	-	noncoding	GE1067	GGAGGCTCTGA GAGTACAGGAG	AGTAGGAACA AAGCCAGGAG	GGAGGCTCTGAGAGTACAGGAGggccccctggatataaactacccctgtcccccaagggtctcgg tggtgtgggctgtggcgcttcttcgctgctcttcttctacgctgctccttctgtgctcaacaaa tgtggacggagaaacaaagtttgggatacaacg[t/g]agtcggggctgcagagggtgtctgtctct gtctgtctTCCCTGGCTTGTTCCTACT
NTRK1u 6	NTRK1	C	T	G	G	cds	GE1129	CAGGCTCCTGG GAGTCTATC	CCAGGTGTCT ACAGTTGGAT	CAGGCTCCTGGGAGTCTATCctccagcctatccccctctcttcttctgttcacagatcccatg gaccogaagccaaagctgtggctgtggggagagtggtggtccag[c/t]ccccctgggtctggg gcagctgtgtggcggtggctagcaggtcgctgcggggatggtgacctggcggtctgcatctttg tgacacgggacctggccacacacgactgtctagtggccaggaggtggtggtcaagattggtgat tttggcatgagcagggatatactacagcacgactattaccgtgaagggtccttgtcccccaacg ccttccccctgcATCCAAACTGTAGACACCCCTGG

FIG. 5FFFFF

[illegible]

[illegible]

[illegible]

FIG. 5

[illegible]

FIG. 5KKKKK

[illegible]

FIG. 5NNNNNN

[illegible]

FIG. 5000000

[illegible]

FIG. 5PPPPP

[illegible]

FIG. 5SSSSS

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' → 3')	Reverse Primer (5' → 3')	Assay Sequence
PCIu5	PCI	C	T	A	V	cds	GE413	GGACATCTCTG GAAAGTCAGC	TGAGGGAATTG GGTATTCTTTA GAT	GGACATCTCTGGAAGTCAGCacttggaacagctccacccctctctgaggaacaccttttccct ttcagaacaaagaaacagccacccatgcagctcttctctctcttggcctgggtgcttctcagccctc agggggcctcccttaccgcccaccccgaggagatgaagaagagagtcgaggaacctccatgta gggtccacgggtggccccccagcagcagaaggaacttacccttgacacctacaggg[c/l]cttgg cttccgtgccccccagcagacatcttcttctccctgtgagcatctccatgagcctggccatg ctctccctgggggtgggtccagcacaaagatgcagatcctggaggcctgggctcaacctcca gaaagctcagagaagagagctgcacagaggtcttcagcagctcttcaggagctcaacacagccca gagatggcttccagctgagcctcggaatgcccccttttccacgacctgggtgagacctgaggac accttcgttaagtgcacatgaagcgtgtacctggcagacacttccccacacctttaggactc tgacggggcccatgaagcagatcaatgattatgtggcaagcaaacggaaggaagatttggaact tgcttaagaacctcgatagcaatgcggtcgatgatggtgaattacatcttctttaaaggtaag gccccctggggcccaacctgcacttcttctggctttctgctgcttttATCTAAGAAATACCCAAT TCCCTCA
PCIu6	PCI	A	G	K	E	cds	GE413	GGACATCTCTG GAAAGTCAGC	TGAGGGAATTG GGTATTCTTTA GAT	GGACATCTCTGGAAGTCAGCacttggaacagctccacccctctctgaggaacaccttttccct ttcagaacaaagaaacagccacccatgcagctcttctctctcttggcctgggtgcttctcagccctc agggggcctcccttaccgcccaccccgaggagatgaagaagagagtcgaggaacctccatgta gggtccacgggtggccccccagcagcagaaggaacttacccttgacacctacagggaccttgcttc gggtccccccagcagcagaatcttcttctccctgtgagcatctccatgagcctggccatgctct ccctgggggtgggtccagcacaaagatgcagatcctggaggcctgggacctcaacctccagaaa agctcagag[a/g]aggagctgcacagaggttccagcagctcttcaggagctcaacctccagccca gagatggcttccagctgagcctcggaatgccccctttccacgacctgggtgagacctcaggac accttcgttaagtgcacatgaagcgtgtacctggcagacacttccccacacctttaggactc tgacggggcccatgaagcagatcaatgattatgtggcaagcaaacggaaggaagatttggaact tgcttaagaacctcgatagcaatgcggtcgatgatggtgaattacatcttctttaaaggtaag gccccctggggcccaacctgcacttcttctggctttctgctgcttttATCTAAGAAATACCCAAT TCCCTCA
PCIu7	PCI	T	A	F	I	cds	GE413	GGACATCTCTG GAAAGTCAGC	TGAGGGAATTG GGTATTCTTTA GAT	GGACATCTCTGGAAGTCAGCacttggaacagctccacccctctctgaggaacaccttttccct ttcagaacaaagaaacagccacccatgcagctcttctctctcttggcctgggtgcttctcagccctc agggggcctcccttaccgcccaccccgaggagatgaagaagagagtcgaggaacctccatgta gggtccacgggtggccccccagcagcagaaggaacttacccttgacacctacagggaccttggttc cggtgccccccagcagcagaatcttcttctccctgtgagcatctccatgagcctggccatgctct ccctgggggtgggtccagcacaaagatgcagatcctggaggcctgggacctcaacctccagaaa agctcagagaagaggtgcacagaggttccagcagctcttcaggagctcaacctccagagaga tggttcagctgagcctcggaatgccccctttccacgacctgggtgagacctgaggaacct tcgttaagtgcacatgaagcgtgtacctggcagacacttccccacacctttaggacttgca ggggcccatgaagcagatcaatgattatgtggcaagcaaacggaaggaagatttggaacttgct taagaacctcgatagcaatgcggtcgatgatggtgaattacatcttctttaaaggtaag gccccctggggcccaacctgcacttcttctggctttctgctgcttttATCTAAGAAATACCCAAT TCCCTCA

FIG. 5TTTTTT

[illegible]

[illegible]

[illegible]

FIG. 5XXXXXX

[illegible]

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
PTHu 12	PTHu	A	G	-	-	noncoding	GE669	GCAATTGACAGAACTAATCAGAAATATGTCGCTTAAAGCAGTACCCCTCCACACACAC	TGTCATTTATTA CCCTCAATCTGT G	GCAATTGACAGAACTAATCAGAAATATGTCGCTTAAAGCAGTACCCCTCCACACACAC ccctgtccctccgaccacacagagggcgctagagcccatctctcttctccaccgtaccccaacac aatctcttaccactctaccacaaataatttcataattcaagcttcagaagctagtagacacatcttcata atttctgggagagtgatcttctcccttactctctcactctcagacacacacacacacacacacacac tcattctcttactgtcttctcacttcaaggagagaaatagagacacacacacacacacacacacac gcagac taattcaaatctcaaatcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttct tttggtttaaatcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttct tttaagttattcaacttcaagagataggtttcttccagctctcacaacacacacacacacacacac aaataattttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttct atccttagctCACAGATTGAGGTAAATGACA	683
PTHu 13	PTHu	T	C	-	-	noncoding	GE679	TTCTTTATCGA TGCAATTTCCAT T	AGAAACATATC CCCTAGATAG A	TTCTTTATCGAATTCATTTGCAATTTGCAATTTGCAATTTGCAATTTGCAATTTGCAATTTG gtgggtttgaaaaaaaggaaggaac tatttaagagtgctcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttct ataggatgatgcttcaaggac cccatcatttaaatagac gggcagtgccac tctaaagggttaac atcttaactgtatataattcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttct ccattgtctcctatataataaactagtcacacacacacacacacacacacacacacacacacac agtaaacattccaaatagac tctactatcactatctcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttct tctactatcactatctcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttct	706
PTHu 14	PTHu	G	T	-	-	noncoding	GE651	GAAACTAACC TGTTTCTCCT TTTC	TTCCTTCTACT AAAAATCTTAA AGGATAG	GAAACTAACCCTGTTTCTCCTTCTTCTTCTTCTTCTTCTTCTTCTTCTTCTTCTTCTTCTTCT agcagagacctcccaaggac atatttattgtctgttaaatctctgttaaatctctgttaaatctctgttaaatctctgttaaatct aaactgcacatttggtcatttggaataatttttttttttttttttttttttttttttttttttttt atttaccataaatt tgaatttctataatt acacaaatgaagtgctctctattttgt gatttctcttctgtgcatgtaaaaaaacacagatattttaaattgttaaaagatgtcttaaaaaat aatcttaattacacatcattgattcagagagaggaattcttcttcttcttcttcttcttcttcttct aatcttaattacacatcattgattcagagagaggaattcttcttcttcttcttcttcttcttcttct	582
PTHu 15	PTHu	C	A	Y	*	cds	GE679	TTCTTTATCGA TGCAATTTCCAT T	AGAAACATATC CCCTAGATAG A	TTCTTTATCGAATTCATTTGCAATTTGCAATTTGCAATTTGCAATTTGCAATTTGCAATTTG tctgtgggtttgaaaaaaaggaaggaacacacacacacacacacacacacacacacacacacac tcatttatttaagagtcctctgttacttcttcttcttcttcttcttcttcttcttcttcttcttct ataggatgatgcttcaaggac cccatcatttaaatagac gggcagtgccac tctaaagggttaac atccttaactgtatataatttataaactagtcacacacacacacacacacacacacacacacacac ccattgtctcctatataataaactagtcacacacacacacacacacacacacacacacacacac agtaaacattccaaatagac tctactatcactatctcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttcttct	706

FIG. 5ZZZZZZ

[illegible]

FIG. 5A

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
SELPd2 4	SELP	C	T	P	L	cds	GE954	TGCCACCCCT GAAGAT	GGGCTGGGGCT GTCCAT	TGCCACCCCTGAAGATtctgaacagggaaacatgatctgccttcattctgcaaaagcattcca gcataagctctagctgcagcttcagttgtgaagaggatttgctattagtgaggac[c/t]ggaagtg gtgcaatgcacagcctcggggtATGGACAGCCCCAGCCC
SELPd2 5	SELP	G	A	V	M	cds	GE446	CTCTGCAGTG AGAGAGTG	CACGTGGAGG CTTATTTG	CTCTGCAGTGAGAGAGTtggagaacttgactcctctcaacac[g/a]tgcctcatgaactgcag ccaacctctgggaaactctcttcttaactgcagctgcagcttcggagctgcagctgcagctgcagag taaatggcccaagctggaatgcttggtcttggaatctggcaAAATAAGCTTCCACAGTG
SELPu1	SELP	A	C	N	T	cds	GE462	TACTCTAGCCA TCAAGTGC	ATATTATTACC TTTGCAGGTG	TACTCTAGCCATCAAGTGCcagaactctttggccacagagcgagctggattgctctgaca ctcgtggagaaatcaaatgttggctccacctgtcattctctgtgaaca[a/c]tggcttaagct ggaggggcccaataatgtggaatgcacaactctctggaagatggtcagctactccacCAACCTGCA AAGGTAAATAAT
SELPu1 0	SELP	A	G	T	A	cds	GE466	TCTCTCAGCT GTGCAGT	AAATCTTACC CTCACAGG	TCTCTCAGCTGTGCAGTgtcagcacctggaaagccccagtgaaagga[a/g]ccatggactgtgt tcatacgtctcactgcttttgctatggtccagctgcagctgcaaaatttgagtgccagccggctacagag tgagggcttggaatgctcgtgctgcaatgactctggacactggtctgcaaccttgccaaCCTGT GAGGTAGGATTT
SELPu1 1	SELP	T	C	T	T	cds	GE462	TACTCTAGCCA TCAAGTGC	ATATTATTACC TTTGCAGGTG	TACTCTAGCCATCAAGTGCcagaactctttggccacagagcgagctggattgctctgaca c[t/c]cgtggagaattcaaatgttggctccacctgtcattctctgtgaacaatggctttaaagct ggaggggcccaataatgtggaatgcacaactctctggaagatggtcagctactccacCAACCTGCA AAGGTAAATAAT
SELPu1 2	SELP	A	G	N	D	cds	GE462	TACTCTAGCCA TCAAGTGC	ATATTATTACC TTTGCAGGTG	TACTCTAGCCATCAAGTGCcagaactctttggccacagagcgagctggattgctctgaca ctcgtggagaatcaaatgttggctccacctgtcattctctgt[a/g]acaatggctttaaagct ggaggggcccaataatgtggaatgcacaactctctggaagatggtcagctactccacCAACCTGCA AAGGTAAATAAT
SELPu1 3	SELP	A	G	T	T	cds	GE421	TTTGTAGCAGG ACCATGA	TCCTATTACC TTTTTGCTCTGA	TTTGTAGCAGGACCATTTGactatccaggaagccttgacttactttggtggagcgttggtctctac [a/g]ataggctcgataaatgggtggagcgtctctggcttctgtaagaaagcgttTCACACAAAA GGTAAATAGGA
SELPu1 4	SELP	G	A	P	P	cds	GE483	AACCAGAAAGA AGTGGCAG	AAGCCCTACC TGCTGTA	AACCAGAAAGATGGCAGatggacttatcattacagcacaaaaagcatactcatggaatatttc ccgtaataactgcagaaatcgctacacagacttagtggccatccagaaataaaaaatgaattgatt acctcaataaggctccctacctactacagctcctactactggtatggatccgaaagacacaataag acatgacatgggtgggaacaaaaggctctcccaagggctgagaaactggctgataatga acctaaacaaaggaacacagagactgctgtagagataacatcaagatcc[g/a]tcagcc cctggcaagtggatgatgagcactgcttgagaaaaagcactgctgtTACACAGGTAGGGC CTT
SELPu1 5	SELP	T	C	F	S	cds	GE466	TCCTCTCAGCT GTGCAGT	AAATCTTACC CTCACAGG	TCTCTCAGCTGTGCAGTgtcagcaccttggaagccccccagtgaaagaaacatggactgtgttcac ccgtcactgctt[t/c]tgctatggctccagctgcaaatttgagtgccagcccggtacagag tgagggcttggaatgctcgtgctgcaatgactctggacactggctcgcaccttgccaaCCTGT GAGGTAGGATTT
SELPu1 6	SELP	C	T	S	F	cds	GE445	ATCCCTTAGCT TTGCAGTG	ACACTCTTACC TTGGCATTC	ATCCCTTAGCTTTCCAGTGcaggaatctcccagttcccaaatgagggccgggtgaactgctcccac cccttcgggtgctctaggtaacagctcgtcagcttcacctgcaatgaggtctgctcctggt ggagacagctgtgctcagtgcttggtacttggaactggaatt[c/t]tgctcctccaGAATGC CAAGGTAAAGAGTGT

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
SELPu17	SELP	C	T	D	D	cds	GE454	TCCACTTAGCT ATTTCGTGTG	ACAGTACCTTG ACAGACTGGG	TCCACTTAGCTATTTCGTGTGAGCGCGTGGAGAGTCTCTGTCACAGGAAGCATGGATTGCTCTCCA TCCTTGAGAGCGTTCAGTATGA[C/T]ACCACTGTAGCTTCGCTGCTGGCTGAAGGTTTCATGCTG TGAGAGGAGCGCGGATATAGTTCGGTGTGATAACTTGGGACAGTGGACAGCACCAGCCCCAGTCTGT CAAGGTACTGT	206
SELPu18	SELP	A	T	S	C	cds	GE446	CTCCTGCAGTG ACAGAGTG	CACGTGGGAGG CTTATTGTG	CTCCTGCAGTGAGAGAGTGTGGAGAACTTGAGTCCCTCAACACGTGCTCATGAACGTGAGCCAC CCTCTGGGAAACTCTCTTTAACTCGCAGTGCAGCTTCCACTGCAGTGCAGCGGTACCAAGTAA TGGGCCCTA/T]GCAAGCTGGAATGCTGGCTTCTGGAACTCTGCAATAAAGCCTCCACAGTG	194
SELPu19	SELP	G	A	S	N	cds	GE466	TCTCTCCAGCT GTGCAGT	AAAATCCTACC CTCACAGG	TCTCTCCAGCTGTGCAGTGTGCAGCACCCTGGAGCCCCCA[G/A]TGAAGGAACCATGGACTGTGT TCATCCGCTCACTGCTTTTGGCTATGGCTGCAGCTGCAATTTGAGTGCAGCGCGGTACAGAG TGAGGGGCTTGGACATGCTCCGCTGCTTGAAGTCTGGACACTGGTCTGCAACCTTGCACACCTGT GAGGTAGGATTTT	209
SELPu20	SELP	T	C	N	N	cds	GE462	TACTCTAGCCA TCAAGTGCC	ATATTATTACC TTTGCAGGTG	TACTCTAGCCATCAAGTGCCAGAACTCTTCCCCAGAGCAGGGCAGCGCTGGATTGTTCTGACA CTCGTGGAGAACTCAATGTTGGCTCCACTGTCTCTCTGTAACTTCTCTTGTAACTTCTGCTTAAAGCT GGAGGGGCCCAATAATGTGGAAATGCACAACTCTCTGGAAATGGTCAAGTCTACTCTCCACCACTGCA AAGGTATAATAT	208
SELPu21	SELP	C	G	S	*	cds	GE483	AACAGAAAGA AGTGGCAG	AAGGCCCTACC TGTGTAA	AACAGAAAGAAGTGGCAGCATGGACTTATCATTAACAGCACAAAAGCATACT[C/G]ATGGAATA TTCCCCGTAATAACTGCCAGAACTGCTACACAGACTAGTGGCCATCCAGAAATAAAATGAAAT GATTACCTCAATAAGGTCTCTACCTACTACAGCTCTCTACTGGATGGGATCCGAAAGAACAA TAAGACATGGACATGGGTGGGAACCAAAAGGCTCTCCAAAGGCTGAGAACTGGGTGATA ATGAACCTAACACAAACAAAGGAAACAAAGGAGTCTGTTGAGATATACATCAAGAGTCCGTCAGCC CCTGGCAAGTGGAAATGATGAGCAGTGTCTGGAAGAAAAGCAGCATTTGTACACAGGTAGGGC CTT	393
SELPu22	SELP	T	A	N	K	cds	GE483	AACAGAAAGA AGTGGCAG	AAGGCCCTACC TGTGTAA	AACAGAAAGAAGTGGCAGCATGGACTTATCATTAACAGCACAAAAGCATACTCATGGAAATTTCT CCGTAATAACTGCCAGAACTGCTACACAGACTAGTGGCCATCCAGAAATAAAATGAAATGATT ACCTCAATAAGGTCTCTACCTACTACAGCTCTCTACTGGATGGGATCCGAAAGAACAA[T/A] TAAGACATGGACATGGGTGGGAACCAAAAGGCTCTCCAAAGGCTGAGAACTGGGTGATA ATGAACCTAACACAAACAAAGGAAACAAAGGAGTCTGTTGAGATATACATCAAGAGTCCGTCAGCC CCTGGCAAGTGGAAATGATGAGCAGTGTCTGGAAGAAAAGCAGCATTTGTACACAGGTAGGGC CTT	393
SELPu23	SELP	T	A	C	S	cds	GE451	TCACAACAGGC ATAGCAT	CCCTGTCATGC TGGAGTT	TCACAACAGGCATAGCATCACTTCTCTCTCCAGGGTGTGCAATGTCCAGCGCTCACCACCTCTGG CAGGGAACCATGTACTGTAGGCGCATCATCCGGGAACCTTTGGTTTAACTACCACCTGTACTTTGG CTGCAACGCTGGATTCAACACTCATAGGAGACAGCACTCTCAGC[T/A]GCAACCTTCCAGGACAA TGGACAGCAGTAACCTCCAGCATGCAGAGG	224
SELPu24	SELP	T	A	N	K	cds	GE462	TACTCTAGCCA TCAAGTGCC	ATATTATTACC TTTGCAGGTG	TACTCTAGCCATCAAGTGCCAGAACTCTTCCCCAGAGCAGGGCAGCGCTGGATTGTTCTGACA CTCGTGGAGAACTCAATGTTGGCTCCACTGTCTCTCTGTAACTTGGCTTAAAGTGGAG TGGGCCCAAT[A]AATGTGGAATGCACAACTCTTGGAAATGGTCAAGTCTACTCTCCACCACTGCA AAGGTATAATAT	208
SELPu25	SELP	T	G	S	A	cds	GE462	TACTCTAGCCA TCAAGTGCC	ATATTATTACC TTTGCAGGTG	TACTCTAGCCATCAAGTGCCAGAACTCTTCCCCAGAGCAGGGCAGCGCTGGATTGTTCTGACA CTCGTGGAGAACTCAATGTTGGCTCCACTGTCTCTCTGTAACTTGGCTTAAAGTGGAG TGGGCCCAATAATGTTGGAATGCACAACTT[A/G]CTGGAAGATGGTCAAGTCTACTCTCCACCACTGCA AAGGTATAATAT	208

FIG. 5DDDDDDDD

[illegible]

[illegible]

FIG. 5FFFFF

[illegible]

SUBSTITUTE SHEET (RULE 26)

[illegible]

FIG. 5

171/178

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
TBXAS1 a16	TBXAS 1	A	G	-	-	noncoding	GE332	CGAGATTGAAA TTTAAGGAAA GAC	CCAGAAACACA AGTGGTAACTG A	CGAGATTGAAAATTTAAGGAAAAGACAAAatgctgtgagatttgggttaacacga[a/g]cttctc ccttctgacgacccctccatcagatgggcccctgagttctgagcctcaggaagggccctgccccta tctggacatggtgattgacagagacgctgaggtgacccgacgcttccaggtgtggtgagcccc cctccctgcccagtcctccctcctacccctgccccagcctgaggtcagggccct cctccatCAGTTACCACTTGTGTTCGG
TBXAS1 a17	TBXAS 1	C	T	-	-	noncoding	GE332	CGAGATTGAAA TTTAAGGAAA GAC	CCAGAAACACA AGTGGTAACTG A	CGAGATTGAAAATTTAAGGAAAAGACAAAatgctgtgagatttgggttaacacgaacttctccctt tgtcagcaccctccat[c/t]agatggcccctgagttctgagcctcaggaagggccctgccccta tctggacatggtgattgacagagacgctgaggtgacccgacgcttccaggtgtggtgagcccc cctccctgcccagtcctccctcctacccctgccccagcctgaggtcagggccct cctccatCAGTTACCACTTGTGTTCGG
TBXAS1 d12	TBXAS 1	C	G	Q	E	cds	GE257	TGCTGTTCCAA ATTGTTTACTG A	TTCAACACGCG AAAATCAAAAT	TGCTGTTCCAAATTGTTTACTGAataagtttgaataattgggaatttggcttaattcttctac tatagtgctgtgttctctcaggggttttttgggaagc[c/g]aaatggagctcagaaagctgtg tgacctctgtgtggtgaagaaggaactcaaccgttctattatgtacgatatcttctattatgt acgatATTGATTTCACGTGTGAA
TBXAS1 d13	TBXAS 1	G	A	R	H	cds	GE326	TGAAACCTAT TCTTTTGCTT T	TACAGGCATGA GCCACTGT	TGAAACCTATTTTTCCTTacttcacagagagctagtaattcttaggttcttaataagagcct aaagcatgagtgcaacttcattctcagcttttgaataatgcttttccctccaggtactccacat cagcatctcagacagtggaagttaggtcctcagacatcccaagccttctcttctcattgggaac ttgacatttttcc[g/a]ccaggttaaggtgtcttccattggcttccataataatgctga gggcccagggcACAGTGGCTCATGGCTGTA
TBXAS1 d14	TBXAS 1	G	A	-	-	noncoding	GE355	CTTGGAGCATC CTTGTCTCA	GCTCTCAGCA GAGAACTGG	CTTGGAGCATCTTGTCTCAGatgcaggggtggctcagctggagcacagggctgcagaggggaggg gaggggtgttctgggacagccctgacacacagcagctgcaggttccaggtgcagggccgagc agcagggcccttcaactcctgcccctcggggccgcccacagagctgctcgggtgcatcta gggtgtgtgaggtcaagttgacactgctccac[g/c]tgctgacaaagtccgggttccaaagcct tgagacccaggtgagggcccccctgctcagagggcag[g/a]tcaggggaggggtgggaggggcca CCCCAGTCTCTGCGTGAGAGC
TBXAS1 u1	TBXAS 1	G	C	V	L	cds	GE355	CTTGGAGCATC CTTGTCTCA	GCTCTCAGCA GAGAACTGG	CTTGGAGCATCTTGTCTCAGatgcaggggtggctcagctggagcacagggctgcagaggggaggg gaggggtgttctgggacagccctgacacacagcagctgcaggttccaggtgcagggccgagc agcagggcccttcaactcctgcccctcggggccgcccacagagctgctcgggtgcatcta gggtgtgtgaggtcaagttgacactgctccac[g/c]tgctgacaaagtccgggttccaaagcct ggcctgagacccaggtgagggcccccctgctcagagggcaggggtgcaggggggcca CCCCAGTCTCTGCGTGAGAGC
TBXAS1 u10	TBXAS 1	A	C	-	-	noncoding	GE249	ATGGACCTGTA TTGCCACCA	GAGAGTTTGGCA TTTCTCATGTC TTA	ATGGACCTGTATTGCCACCAaggtggcttgggtcccctgagctcctgacccctctgctt[a/c] cttcccacagggctgggttggagttcaggtcggtagcagacagcttctgttttctacgtgac aaaagatgggaagaggtcagaggtgcccctgctgtctgtctcagtcctgaaagctgaacgaggt AAGACATGAGAAATGCRAACTCTC
TBXAS1 u11	TBXAS 1	C	T	T	M	cds	GE355	CTTGGAGCATC CTTGTCTCA	GCTCTCAGCA GAGAACTGG	CTTGGAGCATCTTGTCTCAGatgcaggggtggctcagctggagcacagggctgcagaggggaggg gaggggtgttctgggacagccctgacacacagcagctgcaggttccaggtgcagggccgagc agcagggcccttca[c/t]gtacctgcccctcggggccgcccacagagctgctcgggtgca tctaggggtgtgtgaggtcaagttgacactgctccacaggttccaggttccaaagcct ggcctgagacccaggtgagggcccccctgctcagagggcaggggtgcaggggggcca CCCCAGTCTCTGCGTGAGAGC

FIG. 5JJJJJJ

172/178

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence	
TBXAS1 u2	TBXAS 1	G	A	A	T	cds	GE274	CGACCTGGTGT TTCCCTCA	TGCTGCCTCCA CTGGTAAAT	CGACCTGGTGTTCCTCAGattccacgggagggcagctcagactgaggtgctggggagcg catcccgaggc(g/a)ctgtgtagagatggcgtgggtgcccctgacccatgacctgagcac tgcccaagcccgagagaccttcaacctgaaaggtgagtgactgcccccttttaaaagctctgaagg gatgtgagtggtggatagaaATTACAGTGGAGGAGCA	237
TBXAS1 u3	TBXAS 1	C	T	A	A	cds	GE274	CGACCTGGTGT TTCCCTCA	TGCTGCCTCCA CTGGTAAAT	CGACCTGGTGTTCCTCAGattccacgggagggcagctcagactgaggtgctggggcagcg catcccgaggcgtgtgttagagatggc(c/t)gtgggtgcccctgacccatgacctgagcac tgcccaagcccgagagaccttcaacctgaaaggtgagtgactgcccccttttaaaagctctgaagg gatgtgagtggtggatagaaATTACAGTGGAGGAGCA	237
TBXAS1 u4	TBXAS 1	C	G	Q	E	cds	GE274	CGACCTGGTGT TTCCCTCA	TGCTGCCTCCA CTGGTAAAT	CGACCTGGTGTTCCTCAGattccacgggagggcagct(c/g)aggactgaggtgctggggc agcgcaccccgaggcgtgtgttagagatggcgtgggtgcccctgacccatgacctgagcac tgcccaagcccgagagaccttcaacctgaaaggtgagtgactgcccccttttaaaagctctgaagg gatgtgagtggtggatagaaATTACAGTGGAGGAGCA	237
TBXAS1 u5	TBXAS 1	G	A	R	Q	cds	GE355	CTTGGAGCATC CTTGCTCA	GCTCTCAGCA GAGAACTGG	CTTGGAGCATCTTGTCTCAGatgcagggtggctcagctggagcacagggtgcagaggaggg gagcgggtgttctggggcagccctgacacacagcagctgcaggttcacggctgagggccgagc agcaccggcccttcagctccctcgccctggggccgcccagcgtgcccctgggtgcatctta ggcctgcttgaggtcaagttagacactgctccagctgctgcacaggtcc(g/a)gttccaagcct gcccgtgagaccaggtgagggccctgctcagaggaggtacaggggagcgggtggggagggcca ccccaggtCTCTGCTGAGAGC	347
TBXAS1 u6	TBXAS 1	T	G	V	G	cds	GE470	GGCCCTGGTGT ATTATCACC	CCAAAGTCGGC TCCATTC	GGCCCTGGTGTATTATCACCcccttttcaatgccactttgttttcttcaagtatcttct catcataatggtccactggcccgatgttgcacaaataagaacccagagagcgaactgaatggcttt tttaacaaactcataggaatg(t/g)gatgtccttggggagcagcagctgccgaagaggttaa cgtattttaataggacacagccttgaatgGAATGGAGCCGACTTTGG	243
TBXAS1 u7	TBXAS 1	T	C	I	T	cds	GE912	GCCCATGTATC TTCCCTCCTTT	GGGGATCCAA CTTGTAAT	GCCCATGTATCTTCCCTCTTgttctccaggaaagcctcactcttcatgactgtaaggtcaaatg tgcattttctccttttcttctttagggcggagagacttctccaaatggtcctggtgcccga cattctgcaagtccttggcgtgcaagactttgacatcgtcagagagcgtttctcctctactgg gtgcaagccgaaccttccgggcaacaccagccagcctatggccagcctttgactgtggtatg aga(t/c)tggtggccagccttcatcttctcactcgtggtggtatgaatcatcaccacacact tcttttgccacctactggtgcccacccctgactgccaagagagccttctgagagaggttag acgtttttaaaggagaaacacgtgAGTACAAAGTTGGATCCCC	432
TBXAS1 u8	TBXAS 1	C	G	L	V	cds	GE912	GCCCATGTATC TTCCCTCCTTT	GGGGATCCAA CTTGTAAT	GCCCATGTATCTTCCCTCTTgttctccaggaaagcctcactcttcatgactgtaaggtcaaatg tgcattttctccttttcttctttagggcggagagacttctccaaatggtcctggtgcccga cattctgcaagtccttggcgtgcaagactttgacatcgtcagagagcgtttctcctctactgg gtgcaagccgaaccttccgggcaacaccagccagcctatggccagcctttgactgtggtatg aga(t/g)tggtggccagccttcatcttctcactcgtggtggtatgaatcatcaccacacacttct tttgccacctac(c/g)actggccacacacccctgactgccaagagagccttctgagagaggttag acgtttttaaaggagaaacacgtgAGTACAAAGTTGGATCCCC	432
TBXAS1 u9	TBXAS 1	T	G	M	R	cds	GE274	CGACCTGGTGT TTCCCTCA	TGCTGCCTCCA CTGGTAAAT	CGACCTGGTGTTCCTCAGattccacgggagggcagctcagactgaggtgctggggcagcg catcccgaggcgtgtgttagagatg(t/g)ggcgtgggtgcccctgacccatgacctgagcac tgcccaagcccgagagaccttcaacctgaaaggtgagtgactgcccccttttaaaagctctgaagg gatgtgagtggtggatagaaATTACAGTGGAGGAGCA	237

FIG. 5KKKKKKK

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/ noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
THBDu1	THBD	T	G	C	G	cds	GE409	TCGGCTTACAG CTAATGTGC	GCCAGCTAAGG TGCTTTGGT	TCGGCTTACAGCTAATGTCCACCGCGCCCGGAGCGGTCACAGGGGCACTGGGCGGAGGCGG CCGGGCGCTTGGGACTGCAAGCGTGGAGAACGGCGGCTGCGAGCACGCGTGAATCGGATCCCTGG GGTCCCGCTGCCAGTGCACGAGCCGCGCCCTGCCAGGACGACGGGCGCTCCTGCACCGCAT CCGGACGCGAGTCCCTGCAACGACCTCTCGAGCACTTCTGCGTCCCAACCCCGACCGCGGCG TCCTACTGTCATGTGGAGACCGGCTACCGGCTGGCGCGGACCAACACCGGTGCGAGGACGT GGATGACTGATATACTGGAGCCCACTCCGTGTCGCGAGCGGTGTCAACACACAGGGTGGCTTCG AGTGCCACTGCTACCTAACTACGACCTGGTGGACGGGAGT/G)GTGGAGCCCGTGGACCC GTGCTTCAGAGCCAACTGGAGTACCACTGCGAGCCCTGAACCAAACTAGTACCTCTGCTGT GCGCGAGGGGCTTCCGCGCATTCGCCAGCGCGCACAGGTGCGAGTGTGTTGCAACCAAGACT GCCTGTCCAGCGGACTGGACCCCAACACAGGCTAGCTGTGAGTGCCTGGAAGGCTACATCCT GGACGACGGTTCATCTGACGCGGACATCGACGAGTGCAGAAACGGCGCTTCTGCTCCGGGTGT GCCACAACCTCCCGGTACTCTGAGTGCATCTGCGGGCCGAGCTCGGCCCTGCCCGCCACATC GGCAACCGACTGTGACTCCCGCAAGGTGGACGGTGGCGACAGCGGCTCTGGCGAGCCCCCGCCAG CCCGACGCCGGCTCCACTGACTCCTCCGCGCTCCGCGCTGGGTGCGCTTCTGGGCTTGTCTCATAG GCATCTCCATCGCGAGCCTGTGCTGGTGGTGGCGCTTCTGGGCGCTCCTCTGCCACCTGCGCAAG AAGCAGGGCGCGCCCGGCGCAAGATGGAGTACAAGTGGCGGGGCCCCCTT
THBDu2	THBD	C	A	P	T	cds	GE409	TCGGCTTACAG CTAATGTGC	GCCAGCTAAGG TGCTTTGGT	TCGGCTTACAGCTAATGTCCACCGCGCCCGGAGCGGTCACAGGGGCACTGGGCGGAGGCGG CCGGGCGCTTGGGACTGCAAGCGTGGAGAACGGCGGCTGCGAGCACGCGTGAATCGGATCCCTGG GGTCCCGCTGCCAGTGCACGAGCCGCGCCCTGCCAGGACGACGGGCGCTCCTGCACCGCAT CCGGACGCGAGTCCCTGCAACGACCTCTGGAGCACTTCTGCGTCCCAACCCCGACCGCGGCG TCCTACTGTCATGTGGAGACCGGCTACCGGCTGGCGCGCGGACCAACCGGTGCGAGGACGT GGATGACTGATATACTGGAGCCCACTCCGTGTCGCGAGCGGTGTCAACACACAGGGTGGCTTCG AGTGCCACTGCTACCTAACTACGACCTGGTGGACGGGAGTGTGGAGCCCGTGGACCCGTGC TTCAGAGCCAACTGCGAGTACCACTGCGAGTC/a)CCCTGAACCAAACTAGTACCTCTGCTGT GCGCGAGGGGCTTCCGCGCATTCGCCAGGAGCGCACAGGTGCCAGTGTGTTGCAACCAAGACT GCCTGTCCAGCGGACTGGACCCCAACACAGGCTAGCTGTGAGTGCCTGAAGGCTACATCCT GGACGACGGTTCATCTGACGCGGACATCGACGAGTGCAGAAACGGCGCTTCTGCTCCGGGTGT GCCACAACCTCCCGGTACTCTGAGTGCATCTGCGGGCCGAGCTCGGCCCTGCCCGCCACATC GGCAACCGACTGTGACTCCCGCAAGGTGGACGGTGGCGACAGCGGCTCTGGCGAGCCCCCGCCAG CCCGACGCCGGCTCCACTGACTCCTCCGCGCTGGGTGGGTGCGCTTCTGGGCGCTCCTCTGCCACCTGCGCAAG GCATCTCCATCGCGAGCCTGTGCTGGTGGTGGCGCTTCTGGGCGCTCCTCTGCCACCTGCGCAAG AAGCAGGGCGCGCCCGGCGCAAGATGGAGTACAAGTGGCGGGGCCCCCTT

FIG. 5LLLLLLL

Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
THP0d5	THPO	C	?	-	-	noncoding	GE345	TCACACTGCTG GCTACTCTCTAA	CCTTCTTCCT CAGGTCTTCTA	TCACCTCTGCTGGCTACTCTCTAAAGGCTCCCCACCCGCTTTTtagtgcccttttagtgcccttttaggcagtgccgt336 tctctctccatctcttctcagagagagacacagagacacagacattctgggagagtgaccctt ctgtggaggagtgatggcagcagcgggacacactgggacccacttgcctctcatccctcctggg gcagcttctggacaggtccgtctctctcttggggcctgcagagcctctcttggaaacccaggtaa gtccccagtcagggtatctgtagaaactgtctcttctgactcagtcctc[c]CTAGAAGACCTG AGGAAAGAAAGG
THP0u1	THPO	T	A	S	T	cds	GE416	GGCATCTCTGTC TTTCTCTACTTA GAC	AGGAAATCTTG TCCAGTTGTCT C	GGCATCTCTGCTTTCTACTTAGACAaggaggcctgagatctggccctgggtgttggccctcagg844 accatctctgcccctcagcttctccacagggcagacacacagctccacaggtatcccaatgccat cttctcagcttcccaacacctgctccgaggaagggtgcgttctctgagtcttctgtaggggtcca ccctctgctcagggggcccccacacacacagctgtcccgagacacactctctctctcaca ctgaacgagctcccaacacaggaacttctggattgttggagacaaactcactgcc[t/a]cagcca gaactactggctctgggtctctgaagtggcagcagggatctcagagcaaatctctgggtctgctg aaccaacctccaggtccctggacaaatccccggtatccctgaacaggtatcacgaactcttgaa tggaactcgtggactcttctcggacctcagcaggaacctaggagccccggacattctctcag gaacatcagacacaggtctctcgcaccccaacctcagcctggattctctctcccaacacct cctctactggacagatacgtctctctctctccacacaccttcacacacaccttgcccacaccttggtctcagct ccacccctgcttctcagaccttctgctccaaagccacacctaccagccctctctctaaacacat cctacacccactccagaatctgtctcaggaagggttaagggtctcagacactgcgcagacatcagca ttgtctcgtgtacagctcccttccctgcaggggggcccttggGAGACAACCTGGACAAGATTTCCT244
THP0u2	THPO	A	G	Q	R	cds	GE265	TGGAGGACTAG CCTGCTTATTA	AGAATCCATGG GAAGCAGTG	TGGAGGACTAGCCTGCTTATTAGgtaccatagctctctctattttagctcccttctccccccac caattcttctcaacagagcc[a/g]gtgcccagaggttcaaccttgcctacacctgtctcgtg cctggtgtggactttagcttgggagaatggaaaaccagagtggttaagaaggccatccctaaacctt ggcttccctaagtcctgtctctcagttctccACTGCTTCCATGGATTCT
THP0u3	THPO	G	A	G	R	cds	GE416	GGCATCTCTGTC TTTCTCTACTTA GAC	AGGAAATCTTG TCCAGTTGTCT C	GGCATCTCTGCTTTCTACTTAGACAaggaggcctgagatctggccctgggtgttggccctcagg844 accatctctgcccctcagcttctccacagggcagacacacagctccacaggtatcccaatgccat cttctcagcttcccaacacctgctccgaggaagggtgcgttctctgagtcttctgtaggggtcca ccctctgctcagggggcccccacacacacagctgtcccgagacacactctctctagctccctaca ctgaacgagctcccaacacaggaacttctggattgttggagacaaactcactgcctcagccagAAC tactggctctgggtctctgaagtggcagcag[g/a]gattcagagccaaagtctctgggtctgctg aaccaacctccaggtccctggacaaatccccggtatccctgaacaggtatcacgaactcttgaa tggaactcgtggactcttctcggacctcagcaggaacctaggagccccggacattctctcag gaacatcagacacaggtccctgcaccccaacctccagcctggattctctctcccccaacct cctcctactggacagatacgtctctctctctccacacaccttcacacacaccttgcccacaccttggtctcagct ccacccctgcttctcagaccttctgctccaaagccacacctaccagccctctctctaaacacat cctacacccactccagaatctgtctcaggaagggttaagggtctcagacactgcgcagacatcagca ttgtctcgtgtacagctcccttccctgcaggggggcccttggGAGACAACCTGGACAAGATTTCCT

FIG. 5NNNNNNN

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Poly Id	Gene	ref NT	alt NT	ref AA	alt AA	coding/noncoding	Assay #	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Assay Sequence
VLDLRd 13	VLDLR	C	T	A	A	cds	GE926	GAAAGACCTTGG CCTTCTTAAAG C	CAAGTGACAAT GACTTATGTCA AGA	GAAAGACCTTGGCTTCTTAAAGCAaaactaagtaaccacccagacttccatcttgcaggcaagagcc aagtctgatttcaactaataagaagacatacaggaagatggcttagagaggaagaataatataatcc aactagttgaacagctaagaacacactgtggtctcgcagatgctgacatctgctcccaagaactatctc tgggc[c/t]gatctaaagccaaaggctatcttcaggtaacttcagttcccttttgggtgTCTT GACATAAGTCATTGTCACTTG
VLDLRd 14	VLDLR	A	T	-	-	noncoding	GE926	GAAAGACCTTGG CCTTCTTAAAG C	CAAGTGACAAT GACTTATGTCA AGA	GAAAGACCTTGGCTTCTTAAAGCAaaactaagtaaccaccc[a/t]gacttccatcttgcaggcaag agccaaactgatttcaactaataagaagacatacaggaagatggcttagagaggaagaataatataat atccaaactagttgaacagctaagaacacactgtggtctcgcagatgctgacatctgctcccaagaact attctgggcccgcagctaaagccaaaggctatcttcaggtaacttcagttcccttttgggtgTCTT GACATAAGTCATTGTCACTTG
VLDLRd 15	VLDLR	G	T	-	-	noncoding	GE937	AGGTTTGGCT CCTTACC	GGTAGCTCCAG ATGAACAAAA	AGGTTTGGCTTGGCTTACCTgattgggttaaatttctaagtcgtgaatacagatcccttcaaaactgatt ccttttattctctctgtagggaatcaatgtgaccacagcagatcagaggtcagtggttcccccaaaa gggacttctgcgcagatggcccattctctctctctgtaagta[g/t]atttccctannngtctgggt tcaagaacttcttagataccagatgaagattTTTGTTCATCTGAGCTACC
VLDLRd 16	VLDLR	G	A	E	K	cds	GE940	CCTGGGTTTAA AATGTGAAGA TA	TATCCTTTCCC ATCACCTGC	CCTGGGTTTAAATGTGAAGATaatttgaaataaagttgtgaagtgantantacatttttat tccagatataaagaaatgcttgggttaaataataggtggatgttctcagatctgcgaagaccttagtta taggctac[g/a]agtgtagctgtgagctgggtttgaaactgagataggaagaaacctgtggagg tgagtcataagaaagaaacctggacctGCAGGTGATGGAAAGGATA
VLDLRd 17	VLDLR	C	T	N	N	cds	GE953	TTTTCACAGCT TTGTTTACTGG T	TGAAGATAGTT GAGTGGGTGGT	TTTTCACAGCTTTGTTTACTGGTgagactgggtgaaccagctcaaaatagaaagagaggaatga atggattcgtatagactccactgggtgacagcgagatccagtggtctaa[c/t]ggaaattacact tgggtatgatttcttctctctcgcACCCACCTCAACTATCTTCA
VLDLRu 1	VLDLR	A	G	T	A	cds	GE920	GCTCTAATTGT GTCAAACTCTT AAAT	GACCTACACAG ATACCATTTCCA AAG	GCTCTAATTGTGTCAAACTCTTAAATtcttcttgacactattctgttcagtgccctaaattgatga caaggttggtagacatgtttaaataatgatcgacaatgtctataatctgcagccatctgtgttggatt gggtgtacaagacactactactgagatgagctgcttcaagactattcagtagactacccttagat ggga[a/g]cgaagaggaagttctgtttaaactctgtactgagagagccctgcctccatagctgtggtg accacatgtctgggttctgtagctgttcttccatcacagaCTTGGAAATGGTATCTGAGCTAGGTC
VLDLRu 10	VLDLR	A	T	S	C	cds	GE937	AGGTTTGGCT CCTTACC	GGTAGCTCCAG ATGAACAAAA	AGGTTTGGCTTGGCTTACCTgattgggttaaatttctaagtcgtgaatacagatcccttcaaaactgatt ccttttattctctctgtagggaatcaatgtgaccacagcagatcagaggtc[a/t]gtgttcccc aaaagggaacttctgcgcagatggcccattctctctctctgtgaagagatttccctannngtctgggt tcaagaacttcttagataccagatgaagattTTTGTTCATCTGAGCTACC
VLDLRu 11	VLDLR	T	C	I	I	cds	GE937	AGGTTTGGCT CCTTACC	GGTAGCTCCAG ATGAACAAAA	AGGTTTGGCTTGGCTTACCTgattgggttaaatttctaagtcgtgaatacagatcccttcaaaactgatt ccttttattctctctgtagggaatcaatgtgaccacagcagatcagaggtcagtggttcccccaaaa gggacttctgcgcagatggcccatt[c/t]cttctctctgtgaagtagatttccctannngtctgggt tcaagaacttcttagataccagatgaagattTTTGTTCATCTGAGCTACC
VLDLRu 12	VLDLR	A	T	S	C	cds	GE945	TCCAATACTAG ACTTAGCTCAC TT	GACTTACTGCT GGGTACGTG	TCCAATACTAGACTTAGCTACTagctaccctctgatttttcttcaagtcgtctcttagtgatggcag cagtaggtggctactttagtggtgggaatttggaacacacagaacatgaaagacatgaaactttgac aatctctgtgtaacttgaaacacactgaagagacctctccatagacattggtagacac[a/t]gtg ccttctgttggacaCACGTACCCACAGTAAGTC
VLDLRu 2	VLDLR	A	G	K	R	cds	GE926	GAAAGACCTTGG CCTTCTTAAAG C	CAAGTGACAAT GACTTATGTCA AGA	GAAAGACCTTGGCTTCTTAAAGCAaaactaagtaaccacccagacttccatcttgcaggca[a/g]ag agccaaactgatttcaactaataagaagacatacaggaagatggcttagagaggaagaataat atccaaactagttgaacagctaagaacacactgtggtctcgcagatgctgacatctgctcccaagaact attctgggcccgcagctaaagccaaaggctatcttcaggtaacttcagttcccttttgggtgTCTT GACATAAGTCATTGTCACTTG

FIG. 5PPPPPPP

RESULT 2

AAC71304

ID AAC71304 standard; DNA; 318 BP.

XX

AC AAC71304;

XX

DT 09-FEB-2001 (first entry)

XX

DE Single nucleotide polymorphism containing sequence #378.

XX

KW Single nucleotide polymorphism; SNP; human; genetic disease;
KW disease susceptibility; cardiovascular system; endocrine system;
KW neurological system; forensic testing; paternity testing; ds.

XX

OS Homo sapiens.

XX

PN WO200058519-A2. /

XX

PD 05-OCT-2000.

XX

PF 30-MAR-2000; 2000WO-US08440.

XX

PR 31-MAR-1999; 99US-0127248.

XX

PA (WHED) WHITEHEAD INST BIOMEDICAL RES.

PA (AFFY-) AFFYMETRIX INC.

XX

PI Altshuler D, Cargill M, Daley GQ, Ireland JS, Lander ES;

PI Lipshutz RJ, Patil N, Sklar P;

XX

DR WPI; 2000-611722/58.

XX

PT Nucleic acid selected from one of 106 genes comprising single
PT nucleotide polymorphisms, allele-specific oligonucleotides to the genes
PT are useful for phenotypic correlations, forensics, paternity testing,
PT medicine and genetic analysis -

XX

PS Claim 1; Fig 5; 214pp; English.

XX

CC The present invention is concerned with a number of human single
CC nucleotide polymorphisms (SNPs) which the inventors identified in human
CC genes. These SNPs can be used in disease diagnosis and prediction of an
CC individual's susceptibility to disease, in forensic and paternity testing
CC and in genetic mapping. In particular, the SNPs of the invention can be
CC used to diagnose susceptibility to diseases of the cardiovascular,
CC endocrine and neurological systems, such as coronary artery disease,
CC schizophrenia, cancer, autoimmune diseases, Alzheimer's and Parkinson's
CC diseases.

CC Note: The degenerate codon within the sequence represents the position
CC of an SNP, for example the letter S represents a polymorphism where the
CC nucleotide may be C or G.

XX

SQ Sequence 318 BP; 66 A; 106 C; 90 G; 55 T; 1 other;

Query Match 99.8%; Score 200.6; DB 21; Length 318;

Best Local Similarity 99.5%; Pred. No. 1.7e-47;

Matches 200; Conservative 1; Mismatches 0; Indels 0; Gaps 0;

Qy	1	TCCCCAGACAAGGATGACCAGCTGATCTGTGTGAACGAGAACGGCGGCTGTGAGCAGTAC	60
Db	94	TCCCCAGACAAGGATGACCAGCTGATCTGTGTGAACGAGAACGGCGGCTGTGAGCAGTAC	153
Qy	61	TGCAGTGACCACACGGGGACCAAGCGCTCCTGTCGGTGCCACGAGGGGTACTCTCTGCTG	120
Db	154	TGCAGTGACCACACGGGGACCAAGCGCTCCTGTCGGTGCCAYGAGGGGTACTCTCTGCTG	213
Qy	121	GCAGACGGGGTGTCCTGCACACCCACAGGTGACCAGGCTTCATGTCCCAGTCCCAGATGA	180
Db	214	GCAGACGGGGTGTCCTGCACACCCACAGGTGACCAGGCTTCATGTCCCAGTCCCAGATGA	273
Qy	181	CACCAGTCCCTGTCCCCTAG	201
Db	274	CACCAGTCCCTGTCCCCTAG	294

RESULT 1

AAC71295

ID AAC71295 standard; DNA; 266 BP.

XX

AC AAC71295;

XX

DT 09-FEB-2001 (first entry)

XX

DE Single nucleotide polymorphism containing sequence #375.

XX

KW Single nucleotide polymorphism; SNP; human; genetic disease;
KW disease susceptibility; cardiovascular system; endocrine system;
KW neurological system; forensic testing; paternity testing; ds.

XX

OS Homo sapiens.

XX

PN WO200058519-A2.

XX

PD 05-OCT-2000.

XX

PF 30-MAR-2000; 2000WO-US08440.

XX

PR 31-MAR-1999; 99US-0127248.

XX

PA (WHED) WHITEHEAD INST BIOMEDICAL RES.

PA (AFFY-) AFFYMETRIX INC.

XX

PI Altshuler D, Cargill M, Daley GQ, Ireland JS, Lander ES;

PI Lipshutz RJ, Patil N, Sklar P;

XX

DR WPI; 2000-611722/58.

XX

PT Nucleic acid selected from one of 106 genes comprising single
PT nucleotide polymorphisms, allele-specific oligonucleotides to the genes
PT are useful for phenotypic correlations, forensics, paternity testing,
PT medicine and genetic analysis -

XX

PS Claim 1; Fig 5; 214pp; English.

XX

CC The present invention is concerned with a number of human single
CC nucleotide polymorphisms (SNPs) which the inventors identified in human
CC genes. These SNPs can be used in disease diagnosis and prediction of an
CC individual's susceptibility to disease, in forensic and paternity testing
CC and in genetic mapping. In particular, the SNPs of the invention can be
CC used to diagnose susceptibility to diseases of the cardiovascular,
CC endocrine and neurological systems, such as coronary artery disease,
CC schizophrenia, cancer, autoimmune diseases, Alzheimer's and Parkinson's
CC diseases.

CC Note: The degenerate codon within the sequence represents the position
CC of an SNP, for example the letter S represents a polymorphism where the
CC nucleotide may be C or G.

XX

SQ Sequence 266 BP; 48 A; 84 C; 78 G; 55 T; 1 other;

Query Match 99.8%; Score 200.6; DB 21; Length 266;
Best Local Similarity 99.5%; Pred. No. 6.8e-48;

Matches 200; Conservative 1; Mismatches 0; Indels 0; Gaps 0;

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Qy      1 GCAGCACTGCAGAGATTTTCATCATGGTCTCCCAGGCCCTCAGGCTCCTCTGCCTTCTGCT 60
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Db      59 GCAGCACTGCAGAGATTTTCATCATGGTCTCCCAGGCCCTCAGGCTCCTCTGCCTTCTGCT 118

Qy      61 TGGGCTTCAGGGCTGCCTGGCTGCAGGTGCGTCCGGGGAGGTTTTCTCCATAAACTTGGT 120
        |||
Db     119 TGGGCTTCAGGGCTGCCTGGCTGCAGGTGCGTCCRGGGAGGTTTTCTCCATAAACTTGGT 178

Qy     121 GGAAGGGCAGTGGGCAAATCCAGGAGCCAGCCCGGGCTTCCCAAACCCCGCCCTTGCTCC 180
        |||
Db     179 GGAAGGGCAGTGGGCAAATCCAGGAGCCAGCCCGGGCTTCCCAAACCCCGCCCTTGCTCC 238

Qy     181 GGACACCCCCATCCACCAGGA 201
        |||
Db     239 GGACACCCCCATCCACCAGGA 259
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